

Table S1 Phenogram candidate genes and literature references for already known genotype-phenotype correlations.

Disease name	Disease ID	Phenogram candidate genes
Xq28 (MECP2) duplication	D:45	MECP2 ¹ ; BGN ² ; L1CAM; ABCD1; AVPR2; SLC6A8
NF1-microdeletion syndrome	MIM:613675	NF1 ³ ; ATAD5 ⁴
Leri-Weill dyschondrosteosis	MIM:127300	SHOX ^{5,6}
Familial Adenomatous Polyposis	MIM:175100	APC ⁷
WAGR 11p13 deletion syndrome	MIM:194072	PAX6 ⁸ ; WT1 ⁸
Pelizaeus-Merzbacher disease	MIM:312080	PLP1 ⁹ ; GLRA4
Potocki-Shaffer syndrome	MIM:601224	ALX4 ¹⁰ ; EXT2 ¹¹⁻¹³ ; CD82; SLC35C1
Split hand/foot malformation 1	MIM:183600	DLX5 ¹⁴ ; DLX6 ¹⁴
Sotos syndrome	MIM:117550	NSD1 ¹⁵ ; SLC34A1 ¹⁶ ; SNCB; PROP1; B4GALT7; CPLX2
Rubinstein-Taybi syndrome	MIM:180849	CREBBP ^{17,18}
Angelman syndrome	MIM:105830	UBE3A ^{19,20} ; OCA2 ^{19,20} ; GABRB3 ^{21,22} ; SNRPN; NDN; NIPA1; GABRA5
RCAD (renal cysts and diabetes)	MIM:137920	HNF1B ^{23,24} ; ACACA; LHX1
Williams-Beuren syndrome	MIM:194050	ELN ²⁵ ; BAZ1B ²⁶ ; LIMK1 ²⁷⁻²⁹ ; GTF2IRD1 ^{26,30,31} ; STX1A; NCF1; ABHD11; FZD9; CLIP2; MLXIPL; FKBP6
Wolf-Hirschhorn syndrome	MIM:194190	WHSC1 ³² ; FGFR1L ³³ ; FGFR3 ³⁴ ; IDUA; CTBP1; TACC3; PDE6B
Potocki-Lupski syndrome	MIM:610883	RAII ³⁵ ; ULK2; TOM1L2; MYO15A; ALDH3A2; ATPAF2; PEMT; SREBF1; MAPK7; EPN2
9q subtelomeric deletion syndrome	MIM:610253	EHMT1 ³⁶ ; CACNA1B
Phelan-Mcdermid syndrome	MIM:606232	SHANK3 ³⁷ ; ARSA; MAPKBIP2
Prader-Willi syndrome	MIM:176270	GABRB3 ^{22,38-40} ; HERC2 ⁴¹ ; NDN ⁴² ; NIPA1 ⁴³ ; OCA2 ⁴⁴⁻⁴⁶ ; UBE3A; GABRA5; SNRPN
17q21.3 microdeletion syndrome	MIM:610443	MAPT; CRHR1
Miller-Dieker syndrome	MIM:247200	ABR ⁴⁷ ; CRK ^{48,49} ; DPH1 ^{48,49} ; HIC1 ⁵⁰ ; MNT ^{48,49} ; PAFAH1B1 ⁵¹ ; PTPNA ^{48,49} ; YWHAE ⁴⁸ ; SERPINF2 ⁵² ; MYO1C; NXN; RPA1; SERPINF1; SRR; VPS53
15q26 overgrowth syndrome	D:81	IGF1R ⁵³ ; ALDH1A3; PCSK6; CHSY1
1p36 microdeletion syndrome	MIM:607872	SKI ⁵⁴ ; GABRD ^{55,56} ; HES5 ^{55,56} ; DVII ^{55,56} ; GNB1; PEX10; TP73; VWA1; NOC2L; AGRN; PRDM16; PRKCZ
Smith-Magenis syndrome	MIM:182290	RAII ^{57,58} ; SREBF1 ⁵⁹ ; MYO15A ⁶⁰ ; LLGL1 ⁶¹ ; PEMT; ALDH3A2; ATPAF2; TNFRDF13B; FLCN; MAPK7; TOM1L2; ULK2; B9D1
15q24 microdeletion syndrome	MIM:613406	CYP11A1 ⁶² ; CSPG4; PTPN9; CSK; STRA6; CAX5A; MPI; NEIL1
1q21.1 susceptibility locus (TAR)	MIM:274000	ITGA10; TXNIP; HFE2
Cri du Chat syndrome	MIM:123450	SLC9A3; SLC6A3; NKD2; TERT; IRX1; CCT5; SDHA; SLC6A19; MTRR; SLC12A7; NDUFS6
3q29 microduplication syndrome	MIM:611936	DLG1 ⁶³ ; NCBP2

Table S1: Summary of candidate genes and references to known genotype-phenotype associations from the literature. The column Disease ID shows the ID for the Online Mendelian Inheritance in Man database⁶⁴ where available (“MIM:xx”). Otherwise, the DECIPIER ID⁶⁵ is shown as “D:xx”. “Phenogram candidate genes” shows genes identified by our method as candidates for individual phenotypic features of the CNV disorders. Previously known associations are indicated by literature citations.

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Table S2 List of all phenogram results for the 27 CNV disorders as well as references to already known genotype-phenotype associations from the literature

Disease (OMIM ID)	Gene (Entrez Gene ID)	Human Phenotype of Syndrome (HPO ID)	Phenogram match from [organism]	Reference
15q24 microdeletion syndrome (MIM:613406)	COX5A (9377)	Hearing impairment (HP:0000365)	abnormally_disrupted_sensory_perception_of_touch [ZP]	-
15q24 microdeletion syndrome (MIM:613406)	COX5A (9377)	Short palm (HP:0004279)	abnormally_decreased_size_mandibular_arch_skeleton [ZP]	-
15q24 microdeletion syndrome (MIM:613406)	CSK (1445)	Microcephaly (HP:0000252)	megacephaly [MP]	-
15q24 microdeletion syndrome (MIM:613406)	CSK (1445)	Recurrent infections (HP:0002719)	increased_susceptibility_to_bacterial_infection [MP]	-
15q24 microdeletion syndrome (MIM:613406)	CSPG4 (1464)	Dysplastic corpus callosum (HP:0006989)	abnormal_dentate_gyrus_morphology [MP]	-
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Cryptorchidism (HP:0000028)	absent_prostate_gland [MP]	PMID:20678247
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Cryptorchidism (HP:0000028)	abnormal_epididymis_epithelium_morphology [MP]	PMID:20678247
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Cryptorchidism (HP:0000028)	abnormal_vas_deferens_morphology [MP]	PMID:20678247
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Cryptorchidism (HP:0000028)	absent_prostate_gland_anterior_lobe [MP]	PMID:20678247
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Cryptorchidism (HP:0000028)	abnormal_seminiferous_tubule_morphology [MP]	PMID:20678247
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Cryptorchidism (HP:0000028)	abnormal_testis_morphology [MP]	PMID:20678247
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Cryptorchidism (HP:0000028)	abnormal_epididymis_morphology [MP]	PMID:20678247
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Growth hormone deficiency (HP:0000824)	decreased_circulating_corticosterone_level [MP]	PMID:20678247
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Growth hormone deficiency (HP:0000824)	increased_circulating_adrenocorticotropin_level [MP]	PMID:20678247
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Growth hormone deficiency (HP:0000824)	decreased_circulating_aldosterone_level [MP]	PMID:20678247
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Growth hormone deficiency (HP:0000824)	abnormal_circulating_hormone_level [MP]	PMID:20678247
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Hypoplasia of penis (HP:0008736)	absent_scrotum [MP]	-
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Hypoplasia of penis (HP:0008736)	testis_hypoplasia [MP]	-
15q24 microdeletion syndrome (MIM:613406)	CYP11A1 (1583)	Hypospadias (HP:0000047)	absent_bulbourethral_gland [MP]	-
15q24 microdeletion syndrome (MIM:613406)	MPI (4351)	Growth hormone deficiency (HP:0000824)	Hyperinsulinemic_hypoglycemia [HS]	MIM:602579
15q24 microdeletion syndrome (MIM:613406)	MPI (4351)	Muscular hypotonia (HP:0001252)	Muscular_hypotonia [HS]	MIM:602579
15q24 microdeletion syndrome (MIM:613406)	NEIL1 (79661)	Growth hormone deficiency (HP:0000824)	increased_circulating_leptin_level [MP]	-
15q24 microdeletion syndrome (MIM:613406)	NEIL1 (79661)	Growth hormone deficiency (HP:0000824)	increased_circulating_insulin_level [MP]	-
15q24 microdeletion syndrome (MIM:613406)	NEIL1 (79661)	Joint laxity (HP:0001388)	joint_inflammation [MP]	-
15q24 microdeletion syndrome (MIM:613406)	NEIL1 (79661)	Obesity (HP:0001513)	obese [MP]	-
15q24 microdeletion syndrome (MIM:613406)	PTPN9 (5780)	Dysplastic corpus callosum (HP:0006989)	abnormal_brain_white_matter_morphology [MP]	-
15q24 microdeletion syndrome (MIM:613406)	PTPN9 (5780)	Nystagmus (HP:0000639)	tremors [MP]	-
15q24 microdeletion syndrome (MIM:613406)	PTPN9 (5780)	Widely spaced teeth (HP:0000687)	abnormal_tooth_morphology [MP]	-
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Abnormality of the nasal bridge (HP:0000422)	Broad_nasal_bridge [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Abnormality of the outer ear (HP:0000356)	Low-set_ears [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Congenital diaphragmatic hernia (HP:0000776)	Congenital_diaphragmatic_hernia [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Congenital diaphragmatic hernia (HP:0000776)	Diaphragmatic_eversion [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Cryptorchidism (HP:0000028)	Cryptorchidism [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Cryptorchidism (HP:0000028)	Pelvic_kidney [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Downslanted palpebral fissures (HP:0000494)	Blepharophimosis [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	High anterior hairline (HP:0009890)	Abnormality_of_the_hair [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Hypoplastic nasal alae (HP:0000430)	abnormally_deformed_Meckel's_cartilage [ZP]	-
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Hypoplastic nasal alae (HP:0000430)	abnormally_deformed_ceratohyal_cartilage [ZP]	-
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Hypoplastic nasal alae (HP:0000430)	abnormally_deformed_palatoquadrate_cartilage [ZP]	-
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Hypoplastic nasal alae (HP:0000430)	abnormally_decreased_size_ceratobranchial_cartilage [ZP]	-
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Inguinal hernia (HP:0000023)	Inguinal_hernia [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Intellectual disability (HP:0001249)	Intellectual_disability_profound [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Muscular hypotonia (HP:0001252)	Muscular_hypotonia [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Short palm (HP:0004279)	Micrognathia [HS]	MIM:601186
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Widely spaced teeth (HP:0000687)	abnormally_morphology_pharyngeal_arch_3-7 [ZP]	-
15q24 microdeletion syndrome (MIM:613406)	STRA6 (64220)	Widely spaced teeth (HP:0000687)	Right_aortic_arch [HS]	-
15q26 overgrowth syndrome (DECIPHER:81)	ALDH1A3 (220)	Downslanted palpebral fissures (HP:0000494)	abnormal_eyelid_morphology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	ALDH1A3 (220)	Large nose (HP:0000461)	abnormal_nose_morphology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	ALDH1A3 (220)	Large nose (HP:0000461)	abnormal_nasal_cavity_morphology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	ALDH1A3 (220)	Large nose (HP:0000461)	ethmoturbinate_hypoplasia [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	ALDH1A3 (220)	Large nose (HP:0000461)	choanal_atresia [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	ALDH1A3 (220)	Prominent nose (HP:0000448)	abnormal_maxillary_sinus_morphology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	ALDH1A3 (220)	Prominent nose (HP:0000448)	abnormal_nose_morphology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	ALDH1A3 (220)	Prominent nose (HP:0000448)	abnormal_nasal_cavity_morphology [MP]	-

15q26 overgrowth syndrome (DECIPHER:81)	ALDH1A3 (220)	Prominent nose (HP:0000448)	ethmoturbinate_hypoplasia [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	ALDH1A3 (220)	Prominent nose (HP:0000448)	choanal_atresia [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	ALDH1A3 (220)	Prominent nose (HP:0000448)	abnormal_maxillary_sinus_morphology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	CHSY1 (22856)	Abnormality of the pinna (HP:0000377)	abnormally_decreased_size_otic_placode [ZP]	-
15q26 overgrowth syndrome (DECIPHER:81)	CHSY1 (22856)	Low-set ears (HP:0000369)	abnormally_decreased_size_otic_placode [ZP]	-
15q26 overgrowth syndrome (DECIPHER:81)	CHSY1 (22856)	Micrognathia (HP:0000347)	abnormally_protruding_ventral_mandibular_arch [ZP]	-
15q26 overgrowth syndrome (DECIPHER:81)	CHSY1 (22856)	Micrognathia (HP:0000347)	abnormally_decreased_size_mandibular_arch_skeleton [ZP]	-
15q26 overgrowth syndrome (DECIPHER:81)	CHSY1 (22856)	Overgrowth (HP:0001548)	abnormally_decreased_length_whole_organism [ZP]	-
15q26 overgrowth syndrome (DECIPHER:81)	CHSY1 (22856)	Sensorineural hearing impairment (HP:0000407)	abnormally_morphology_pars_superior_ear [ZP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Arachnodactyly (HP:0001166)	abnormal Hindlimb_morphology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Arachnodactyly (HP:0001166)	abnormal_calcaneum_morphology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Broad nasal bridge (HP:0000431)	Broad_nasal_bridge [HS]	MIM:270450
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Camptodactyly (hands) (HP:0100490)	Radial_deivation_of_finger [HS]	MIM:270450
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Craniosynostosis (HP:0001363)	abnormal_bone_ossification [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Craniosynostosis (HP:0001363)	delayed_bone_ossification [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Craniosynostosis (HP:0001363)	abnormal_neurocranium_morphology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Craniosynostosis (HP:0001363)	abnormal_bone_mineralization [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Joint hypermobility (HP:0001382)	abnormal_osteoclast_physiology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Joint hypermobility (HP:0001382)	abnormal_osteoblast_physiology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Joint hypermobility (HP:0001382)	abnormal_skeleton_physiology [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Long face (HP:0000276)	Facial_dysmorphism [HS]	MIM:270450
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Long philtrum (HP:0000343)	Long_smooth_philtrum [HS]	MIM:270450
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Long philtrum (HP:0000343)	Thin_upper_lip_vermillion [HS]	MIM:270450
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Macrocephaly (HP:0000256)	Microcephaly [HS]	MIM:270450
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Overgrowth (HP:0001548)	increased_body_length [MP]	PMID:12404101
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Overgrowth (HP:0001548)	increased_body_weight [MP]	PMID:12404101
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Overgrowth (HP:0001548)	Short_stature [HS]	PMID:12404101
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Overgrowth (HP:0001548)	abnormally_decreased_length_whole_organism [ZP]	PMID:12404101
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Sensorineural hearing impairment (HP:0000407)	abnormally_aplastic_posterior_crista_primumdium [ZP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Sensorineural hearing impairment (HP:0000407)	abnormally_aplastic_lateral_crista_primumdium [ZP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Sensorineural hearing impairment (HP:0000407)	abnormally_absent_inner_ear_hair_cell [ZP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Sensorineural hearing impairment (HP:0000407)	abnormally_aplastic_anterior_crista_primumdium [ZP]	-
15q26 overgrowth syndrome (DECIPHER:81)	IGF1R (3480)	Triangular face (HP:0000325)	Facial_dysmorphism [HS]	MIM:270450
15q26 overgrowth syndrome (DECIPHER:81)	PCSK6 (5046)	Large nose (HP:0000461)	absent_nasal_capsule [MP]	-
15q26 overgrowth syndrome (DECIPHER:81)	PCSK6 (5046)	Prominent nose (HP:0000448)	absent_nasal_capsule [MP]	-
17q21.3 microdeletion syndrome (MIM:610443)	CRHR1 (1394)	Conspicuously happy disposition (HP:0100024)	abnormal_anxiety-related_response [MP]	-
17q21.3 microdeletion syndrome (MIM:610443)	CRHR1 (1394)	Conspicuously happy disposition (HP:0100024)	decreased_anxiety-related_response [MP]	-
17q21.3 microdeletion syndrome (MIM:610443)	CRHR1 (1394)	Conspicuously happy disposition (HP:0100024)	increased_anxiety-related_response [MP]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Abnormality of hair pigmentation (HP:0009887)	disheveled_coat [MP]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Abnormality of hair texture (HP:0010719)	disheveled_coat [MP]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Blepharophimosis (HP:0000581)	Eyelid_apraxia [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Conspicuously happy disposition (HP:0100024)	decreased_anxiety-related_response [MP]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Conspicuously happy disposition (HP:0100024)	increased_anxiety-related_response [MP]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Conspicuously happy disposition (HP:0100024)	Inappropriate_sexual_behavior [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Conspicuously happy disposition (HP:0100024)	Mutism [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Conspicuously happy disposition (HP:0100024)	Lack_of_motivation [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Conspicuously happy disposition (HP:0100024)	Inappropriate_laughter [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Conspicuously happy disposition (HP:0100024)	Irritability [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Conspicuously happy disposition (HP:0100024)	Apathy [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Conspicuously happy disposition (HP:0100024)	Echolalia [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Conspicuously happy disposition (HP:0100024)	Disinhibition [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Dislocated hips (HP:0002827)	abnormal_sciatic_nerve_morphology [MP]	MIM:601104
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Epicantus (HP:0000286)	Eyelid_apraxia [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Feeding difficulties (HP:0002022)	Dysphagia [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Generalized hypotonia (HP:0001290)	Limb_dystonia [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Generalized hypotonia (HP:0001290)	Axial_dystonia [HS]	-

17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Generalized hypotonia (HP:0001290)	Rigidity [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Hip dysplasia (HP:0001385)	abnormal_sciatic_nerve_morphology [MP]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Hypermetropia (HP:0000540)	Photophobia [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Hypermetropia (HP:0000540)	Blurred_vision [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Hypermetropia (HP:0000540)	Diplopia [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Positional foot deformities (HP:0005656)	abnormal_sciatic_nerve_morphology [MP]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Ptosis (HP:0000508)	Eyelid_apraxia [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Scoliosis (HP:0002650)	Kyphoscoliosis [HS]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Seizures (HP:0001250)	decreased_susceptibility_to_pharmacologically_induced_seizures [MP]	-
17q21.3 microdeletion syndrome (MIM:610443)	MAPT (4137)	Upturned palpebral fissure (HP:0000582)	Eyelid_apraxia [HS]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Asymmetry of the ears (HP:0010722)	abnormally_decreased_size_otic_vesicle [ZP]	MIM:254300
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Blepharophimosis (HP:0000581)	Ptosis [HS]	MIM:254300
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Campiodactyly (hands) (HP:0100490)	campiodactyly [MP]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Clinodactyly of the 5th finger (HP:0004209)	campiodactyly [MP]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Downslanted palpebral fissures (HP:0000494)	Ptosis [HS]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Epicanthus (HP:0000286)	Ptosis [HS]	MIM:254300
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Flat nose (HP:0000457)	abnormally_morphology_sensory_neuron [ZP]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Flattened nasal bridge (HP:0000425)	abnormally_morphology_sensory_neuron [ZP]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Hypoplastic/small 5th finger (HP:0009237)	campiodactyly [MP]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Low-set ears (HP:0000369)	abnormally_decreased_size_otic_vesicle [ZP]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Microtia (HP:0008551)	abnormally_decreased_size_otic_vesicle [ZP]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Posteriorly rotated ears (HP:0000358)	abnormally_decreased_size_otic_vesicle [ZP]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Small feet (HP:0001764)	abnormal_hindlimb_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Small feet (HP:0001764)	paraparesis [MP]	-
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Thickened helices (HP:0000391)	abnormally_decreased_size_otic_vesicle [ZP]	MIM:254300
1p36 microdeletion syndrome (MIM:607872)	AGRN (375790)	Upturned palpebral fissure (HP:0000582)	Ptosis [HS]	-
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Impaired social interactions (HP:0000735)	abnormal_social/conspecific_interaction [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Impaired social interactions (HP:0000735)	abnormal_nest_building_behavior [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Impaired social interactions (HP:0000735)	abnormal_whisker_trimming_behavior [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Impaired social interactions (HP:0000735)	abnormal_huddling_behavior [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Patent foramen ovale (HP:0001655)	persistent_truncus_arteriosus [MP]	-
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Self-mutilation (HP:0000742)	abnormal_social/conspecific_interaction [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Self-mutilation (HP:0000742)	abnormal_nest_building_behavior [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Self-mutilation (HP:0000742)	abnormal_whisker_trimming_behavior [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Sensorineural hearing impairment (HP:0000407)	increased_cochlear_hair_cell_number [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Sensorineural hearing impairment (HP:0000407)	abnormal_cochlear_inner_hair_cell_morphology [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Sensorineural hearing impairment (HP:0000407)	abnormal_cochlea_morphology [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Sensorineural hearing impairment (HP:0000407)	abnormal_organ_of_Corti_morphology [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Sensorineural hearing impairment (HP:0000407)	abnormal_orientation_of_outer_hair_cell_stereociliary_bundles [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	DVL1 (1855)	Ventricular septal defect (HP:0001629)	persistent_truncus_arteriosus [MP]	-
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Impaired social interactions (HP:0000735)	abnormal_pup_retrieval [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Impaired social interactions (HP:0000735)	pup_cannibalization [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Impaired social interactions (HP:0000735)	abnormal_nest_building_behavior [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Impaired social interactions (HP:0000735)	abnormal_maternal_nurturing [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Oppositional defiant disorder (HP:0010865)	abnormal_depression-related_behavior [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Oppositional defiant disorder (HP:0010865)	decreased_anxiety-related_response [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Oppositional defiant disorder (HP:0010865)	increased_anxiety-related_response [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Oppositional defiant disorder (HP:0010865)	behavioral_despair [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Seizures (HP:0001250)	absence_seizures [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Self-mutilation (HP:0000742)	abnormal_depression-related_behavior [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Self-mutilation (HP:0000742)	decreased_anxiety-related_response [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Self-mutilation (HP:0000742)	increased_anxiety-related_response [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Self-mutilation (HP:0000742)	abnormal_pup_retrieval [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Self-mutilation (HP:0000742)	pup_cannibalization [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Self-mutilation (HP:0000742)	abnormal_nest_building_behavior [MP]	PMID:12119096

1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Self-mutilation (HP:0000742)	abnormal_maternal_nurturing [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GABRD (2563)	Self-mutilation (HP:0000742)	behavioral_despair [MP]	PMID:12119096
1p36 microdeletion syndrome (MIM:607872)	GNB1 (2782)	Cerebral cortical atrophy (HP:0002120)	thin_cerebral_cortex [MP]	-
1p36 microdeletion syndrome (MIM:607872)	GNB1 (2782)	Dysphagia (HP:0002015)	abnormal_suckling_behavior [MP]	-
1p36 microdeletion syndrome (MIM:607872)	GNB1 (2782)	Feeding problems in infancy (HP:0008872)	abnormal_suckling_behavior [MP]	-
1p36 microdeletion syndrome (MIM:607872)	HES5 (388585)	Sensorineural hearing impairment (HP:0000407)	increased Cochlear_outer_hair_cell_number [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	HES5 (388585)	Sensorineural hearing impairment (HP:0000407)	increased_vestibular_hair_cell_number [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	HES5 (388585)	Sensorineural hearing impairment (HP:0000407)	abnormal_uricle_morphology [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	HES5 (388585)	Sensorineural hearing impairment (HP:0000407)	increased_cochlear_inner_hair_cell_number [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	HES5 (388585)	Sensorineural hearing impairment (HP:0000407)	abnormal_vestibular_saccule_morphology [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	HES5 (388585)	Sensorineural hearing impairment (HP:0000407)	abnormal_vestibule_morphology [MP]	PMID:20034100
1p36 microdeletion syndrome (MIM:607872)	NOC2L (26155)	Brachycephaly (HP:0000248)	abnormally_decreased_size_head [ZP]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Brachycephaly (HP:0000248)	Macrocephaly [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Brachycephaly (HP:0000248)	Dolichocephaly [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Brachycephaly (HP:0000248)	Flat_occiput [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Brachycephaly (HP:0000248)	Brachyturricephaly [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Camptodactyly (hands) (HP:0100490)	Single_transverse_palmar_crease [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Camptodactyly (hands) (HP:0100490)	Cubitus_valgus [HS]	MIM:202370
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Clinodactyly of the 5th finger (HP:0004209)	Single_transverse_palmar_crease [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Clinodactyly of the 5th finger (HP:0004209)	Ulnar_deviation_of_the_hand_or_of_fingers_of_the_hand [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Conductive hearing impairment (HP:0000405)	Nystagmus [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Congenital hypothyroidism (HP:0000851)	Redundant_neck_skin [HS]	MIM:214100
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Delayed closure of the anterior fontanelle (HP:0001476)	Stippling_of_the_epiphyses [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Delayed skeletal maturation (HP:0002750)	Delayed_skeletal_maturation [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Dilatation of lateral cerebral ventricles (HP:0006796)	Subependymal_cysts [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Dysphagia (HP:0002015)	Redundant_neck_skin [HS]	MIM:214100
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Epicanthus (HP:0000286)	Epicanthus [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Flat nose (HP:0000457)	Nares_anterverted [HS]	MIM:214100
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Flattened nasal bridge (HP:0000425)	Broad_nasal_bridge [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Flattened nasal bridge (HP:0000425)	Nares_anterverted [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Frontal bossing (HP:0002007)	Dolichocephaly [HS]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Frontal bossing (HP:0002007)	Flat_occiput [HS]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Frontal bossing (HP:0002007)	Frontal_bossing [HS]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Frontal bossing (HP:0002007)	High_forehead [HS]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Gastroesophageal reflux (HP:0002020)	Redundant_neck_skin [HS]	MIM:214100
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Hypertelorism (HP:0000316)	Hypertelorism [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Hypoplastic/small 5th finger (HP:0009237)	Single_transverse_palmar_crease [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Hypoplastic/small 5th finger (HP:0009237)	Micrognathia [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Intellectual disability (HP:0001249)	Intellectual_disability_progressive [HS]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Leukoencephalopathy (HP:0002352)	Aplasia/Hypoplasia_of_the_corpus_callosum [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Low-set ears (HP:0000369)	Low_set_ears [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Malar hypoplasia (HP:0000272)	Malar_hypoplasia [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Malar hypoplasia (HP:0000272)	Round_face [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Microcephaly (HP:0000252)	Macrocephaly [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Muscular hypotonia (HP:0001252)	Muscular_hypotonia [HS]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Patient ductus arteriosus (HP:0001643)	Patent_ductus_arteriosus [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Polymicrogyria (HP:0002126)	Polymicrogyria [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Polymicrogyria (HP:0002126)	Heterotopia [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Posteriorly rotated ears (HP:0000358)	Posteriorly_rotated_ears [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Seizures (HP:0001250)	Seizures [HS]	MIM:202370
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Sensorineural hearing impairment (HP:0000407)	Sensorineural_hearing_impairment [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Small feet (HP:0001764)	Talipes_equinovarus [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Small feet (HP:0001764)	Metatarsus_adductus [HS]	MIM:214100
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Small feet (HP:0001764)	Vertical_talus [HS]	MIM:214100
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Strabismus (HP:0000486)	Esotropia [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Thickened helices (HP:0000391)	Abnormality_of_the_helix [HS]	-

1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Upstalanted palpebral fissure (HP:0000582)	Upstalanted_palpebral_fissure [HS]	MIM:214100
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Ventricular septal defect (HP:0001629)	Ventricular_septal_defect [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PEX10 (5192)	Wide anterior fontanel (HP:0000260)	Widely_patent_fontanelles_and_sutures [HS]	-
1p36 microdeletion syndrome (MIM:607872)	PRDM16 (63976)	Dilated cardiomyopathy (HP:0001644)	ventricular_hypoplasia [MP]	-
1p36 microdeletion syndrome (MIM:607872)	PRDM16 (63976)	Flat nose (HP:0000457)	abnormal_snout_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	PRDM16 (63976)	Flat nose (HP:0000457)	short_snout [MP]	-
1p36 microdeletion syndrome (MIM:607872)	PRDM16 (63976)	Flat nose (HP:0000457)	abnormal_nasal_bone_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	PRDM16 (63976)	Flattened nasal bridge (HP:0000425)	short_nasal_bone [MP]	-
1p36 microdeletion syndrome (MIM:607872)	PRDM16 (63976)	Flattened nasal bridge (HP:0000425)	abnormal_nasal_bone_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	PRDM16 (63976)	Hypoplastic/small 5th finger (HP:0009237)	mandible_hypoplasia [MP]	-
1p36 microdeletion syndrome (MIM:607872)	PRDM16 (63976)	Hypoplastic/small 5th finger (HP:0009237)	small_maxilla [MP]	-
1p36 microdeletion syndrome (MIM:607872)	PRDM16 (63976)	Ventricular septal defect (HP:0001629)	abnormal_heart_ventricle_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	PRKCZ (5590)	Polymicrogyria (HP:0002126)	abnormally_behavioral_quality_of_a_process_cell_migration [ZP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Blepharophimosis (HP:0000581)	eyelids_open_at_birth [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Camptodactyly (hands) (HP:0100490)	polydactyly [MP]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Clinodactyly of the 5th finger (HP:0004209)	polydactyly [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Congenital hypothyroidism (HP:0000851)	abnormal_cervical_vertebrae_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Congenital hypothyroidism (HP:0000851)	abnormal_cervical_atlas_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Congenital hypothyroidism (HP:0000851)	abnormal_cervical_axis_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Downslanted palpebral fissures (HP:0000494)	eyelids_open_at_birth [MP]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Dysphagia (HP:0002015)	abnormal_cervical_vertebrae_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Dysphagia (HP:0002015)	abnormal_cervical_atlas_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Dysphagia (HP:0002015)	abnormal_cervical_axis_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Epicanthus (HP:0000286)	eyelids_open_at_birth [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Gastroesophageal reflux (HP:0002020)	abnormal_cervical_vertebrae_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Gastroesophageal reflux (HP:0002020)	abnormal_cervical_atlas_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Gastroesophageal reflux (HP:0002020)	abnormal_cervical_axis_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Hypoplastic/small 5th finger (HP:0009237)	polydactyly [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Hypoplastic/small 5th finger (HP:0009237)	short_mandible [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Malar hypoplasia (HP:0000272)	midline_facial_cleft [MP]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Scoliosis (HP:0002650)	abnormal_spine_curvature [MP]	-
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Upstalanted palpebral fissure (HP:0000582)	eyelids_open_at_birth [MP]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Wide anterior fontanel (HP:0000260)	abnormal_neurocranium_morphology [MP]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Wide anterior fontanel (HP:0000260)	abnormal_presphenoid_bone_morphology [MP]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Wide anterior fontanel (HP:0000260)	small_basisphenoid_bone [MP]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Wide anterior fontanel (HP:0000260)	small_basiooccipital_bone [MP]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Wide anterior fontanel (HP:0000260)	absent_frontal_bone [MP]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Wide anterior fontanel (HP:0000260)	absent_interparietal_bone [MP]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	SKI (6497)	Wide anterior fontanel (HP:0000260)	absent_parietal_bone [MP]	PMID:11731796
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Brachycephaly (HP:0000248)	domed_craniun [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Cerebral cortical atrophy (HP:0002120)	abnormal_hippocampus_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Cerebral cortical atrophy (HP:0002120)	abnormal_dentate_gyrus_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Cerebral cortical atrophy (HP:0002120)	abnormal_hippocampus_development [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Cerebral cortical atrophy (HP:0002120)	abnormal_cerebral_cortex_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Conductive hearing impairment (HP:0000405)	increased_susceptibility_to_otitis_media [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Dilatation of lateral cerebral ventricles (HP:0006796)	intraventricular_hemorrhage [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Dilatation of lateral cerebral ventricles (HP:0006796)	non-obstructive_hydrocephaly [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Dilatation of lateral cerebral ventricles (HP:0006796)	enlarged_lateral_ventricles [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Dilatation of lateral cerebral ventricles (HP:0006796)	hydroencephaly [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Flat nose (HP:0000457)	rhinitis [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Flat nose (HP:0000457)	abnormal_vomeronasal_organ_morphology [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Flattened nasal bridge (HP:0000425)	rhinitis [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Frontal bossing (HP:0002007)	domed_craniun [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Impaired social interactions (HP:0000735)	submission_towards_male_mice [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Impaired social interactions (HP:0000735)	reduced_male_mating_frequency [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Malar hypoplasia (HP:0000272)	periorbital_edema [MP]	-

1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Polymicrogyria (HP:0002126)	increased_neuron_apoptosis [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Self-mutilation (HP:0000742)	reduced_male_mating_frequency [MP]	-
1p36 microdeletion syndrome (MIM:607872)	TP73 (7161)	Sensorineural hearing impairment (HP:0000407)	abnormally_decreased_size_inner_ear [ZP]	-
1p36 microdeletion syndrome (MIM:607872)	VWA1 (64856)	Conductive hearing impairment (HP:0000405)	increased_thermal_nociceptive_threshold [MP]	-
1p36 microdeletion syndrome (MIM:607872)	VWA1 (64856)	Sensorineural hearing impairment (HP:0000407)	increased_thermal_nociceptive_threshold [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	HFE2 (148738)	Anemia (HP:0001903)	decreased_mean_corpuscular_hemoglobin_concentration [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	HFE2 (148738)	Anemia (HP:0001903)	increased_red_blood_cell_distribution_width [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	HFE2 (148738)	Anemia (HP:0001903)	decreased_mean_corpuscular_volume [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	HFE2 (148738)	Anemia (HP:0001903)	microcytic_anemia [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	HFE2 (148738)	Anemia (HP:0001903)	decreased_hemoglobin_content [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	HFE2 (148738)	Eosinophilia (HP:0001880)	abnormal_macrophage_morphology [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	HFE2 (148738)	Thrombocytopenia (HP:0001873)	decreased_hematocrit [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	HFE2 (148738)	Thrombocytopenia (HP:0001873)	decreased_erythrocyte_cell_number [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	ITGA10 (8515)	Coxa valga (HP:0002673)	short_femur [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	ITGA10 (8515)	Coxa valga (HP:0002673)	abnormal_long_bone_epiphyseal_plate_morphology [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	ITGA10 (8515)	Coxa valga (HP:0002673)	decreased_width_of_hypertrophic_chondrocyte_zone [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	ITGA10 (8515)	Patellar aplasia (HP:0006443)	short_tibia [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	TXNIP (10628)	Anemia (HP:0001903)	hematuria [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	TXNIP (10628)	Eosinophilia (HP:0001880)	increased_lymphocyte_cell_number [MP]	-
1q21.1 susceptibility locus (TAR) (MIM:274000)	TXNIP (10628)	Eosinophilia (HP:0001880)	increased_regulatory_T_cell_number [MP]	-
3q29 microduplication syndrome (MIM:611936)	DLG1 (1739)	Nasal hypoplasia (HP:0003196)	short_snout [MP]	PMID:18241066
3q29 microduplication syndrome (MIM:611936)	NCBP2 (22916)	Macrocephaly (HP:0000256)	abnormally_decreased_size_head [ZP]	-
3q29 microduplication syndrome (MIM:611936)	NCBP2 (22916)	Microcephaly (HP:0000252)	abnormally_decreased_size_head [ZP]	-
9q subtelomeric deletion syndrome	CACNA1B (774)	Aggressive behavior (HP:0000718)	decreased_anxiety-related_response [MP]	-
9q subtelomeric deletion syndrome	CACNA1B (774)	Aggressive behavior (HP:0000718)	increased_aggression_towards_mice [MP]	-
9q subtelomeric deletion syndrome	CACNA1B (774)	Autism (HP:0000717)	decreased_anxiety-related_response [MP]	-
9q subtelomeric deletion syndrome	CACNA1B (774)	Sleep disturbances (HP:0002360)	abnormal_sleep_pattern [MP]	-
9q subtelomeric deletion syndrome	CACNA1B (774)	Sleep disturbances (HP:0002360)	fragmentation_of_sleep_wake_states [MP]	-
9q subtelomeric deletion syndrome	CACNA1B (774)	Sleep disturbances (HP:0002360)	abnormal_frequency_of_paradoxical_sleep [MP]	-
9q subtelomeric deletion syndrome	EHMT1 (79813)	Aggressive behavior (HP:0000718)	abnormal_response_to_novel_object [MP]	PMID:19264732
9q subtelomeric deletion syndrome	EHMT1 (79813)	Aggressive behavior (HP:0000718)	decreased_anxiety-related_response [MP]	PMID:19264732
9q subtelomeric deletion syndrome	EHMT1 (79813)	Aggressive behavior (HP:0000718)	increased_anxiety-related_response [MP]	PMID:19264732
9q subtelomeric deletion syndrome	EHMT1 (79813)	Aggressive behavior (HP:0000718)	abnormal_social_investigation [MP]	PMID:19264732
9q subtelomeric deletion syndrome	EHMT1 (79813)	Aggressive behavior (HP:0000718)	abnormal_social/conspecific_interaction [MP]	PMID:19264732
9q subtelomeric deletion syndrome	EHMT1 (79813)	Aggressive behavior (HP:0000718)	abnormal_response_to_novelty [MP]	PMID:19264732
9q subtelomeric deletion syndrome	EHMT1 (79813)	Aggressive behavior (HP:0000718)	decreased_exploration_in_new_environment [MP]	PMID:19264732
9q subtelomeric deletion syndrome	EHMT1 (79813)	Autism (HP:0000717)	abnormal_response_to_novel_object [MP]	PMID:15805155
9q subtelomeric deletion syndrome	EHMT1 (79813)	Autism (HP:0000717)	decreased_anxiety-related_response [MP]	PMID:15805155
9q subtelomeric deletion syndrome	EHMT1 (79813)	Autism (HP:0000717)	increased_anxiety-related_response [MP]	PMID:15805155
9q subtelomeric deletion syndrome	EHMT1 (79813)	Autism (HP:0000717)	abnormal_response_to_novelty [MP]	PMID:15805155
9q subtelomeric deletion syndrome	EHMT1 (79813)	Autism (HP:0000717)	decreased_exploration_in_new_environment [MP]	PMID:15805155
9q subtelomeric deletion syndrome	EHMT1 (79813)	Obesity (HP:0001513)	obese [MP]	PMID:15805155
Angelman syndrome (MIM:105830)	GABRA5 (2558)	Cerebral cortical atrophy (HP:0002120)	abnormal_hippocampus_CA1_region_morphology [MP]	-
Angelman syndrome (MIM:105830)	GABRA5 (2558)	Hyperactivity (HP:0000752)	hyperactivity [MP]	PMID:9514592
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Hyperactivity (HP:0000752)	hyperactivity [MP]	PMID:9108119
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Paroxysmal bursts of laughter (HP:0000749)	abnormal_parental_behavior [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Paroxysmal bursts of laughter (HP:0000749)	abnormal_maternal_nursuring [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Paroxysmal bursts of laughter (HP:0000749)	reduced_male_mating_frequency [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Paroxysmal bursts of laughter (HP:0000749)	abnormal_response_to_novel_object [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Paroxysmal bursts of laughter (HP:0000749)	abnormal_social/conspecific_interaction [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Paroxysmal bursts of laughter (HP:0000749)	abnormal_nest_building_behavior [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Progressive gait ataxia (HP:0007240)	abnormal_cerebellum_vermis_lobule_V_morphology [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Progressive gait ataxia (HP:0007240)	abnormal_cerebellum_vermis_lobule_VI_morphology [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Progressive gait ataxia (HP:0007240)	abnormal_cerebellum_vermis_lobule_VII_morphology [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Progressive gait ataxia (HP:0007240)	abnormal_cerebellum_vermis_lobule_II_morphology [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Progressive gait ataxia (HP:0007240)	abnormal_cerebellum_vermis_lobule_III_morphology [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Progressive gait ataxia (HP:0007240)	abnormal_cerebellum_vermis_lobule_IV_morphology [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Progressive gait ataxia (HP:0007240)	abnormal_cerebellum_vermis_lobule_morphology [MP]	-
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Seizures (HP:0001250)	seizures [MP]	PMID:9108119
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Seizures (HP:0001250)	absence_seizures [MP]	PMID:9108119
Angelman syndrome (MIM:105830)	GABRB3 (2562)	Absent speech development (HP:0001344)	Impaired_language_development [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Blond hair (HP:0002214)	Frontal_upsweep_of_hair [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Blond hair (HP:0002214)	Hypopigmentation_of_hair [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Blue irides (HP:0000635)	Reduced_iris_pigmentation [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Flat occiput (HP:0005469)	Dolichocephaly [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Hyperactivity (HP:0000752)	Attention_deficit_hyperactivity_disorder [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Hypopigmentation of the skin (HP:0001010)	Hypopigmentation_of_the_skin [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Hypoplasia of the maxilla (HP:0000327)	Short_palm [HS]	-
Angelman syndrome (MIM:105830)	NDN (4692)	Microbrachycephaly (HP:0002258)	Dolichocephaly [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Motor delay (HP:0001270)	Motor_delay [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Muscular hypotonia (HP:0001252)	hypotonia [MP]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Muscular hypotonia (HP:0001252)	Generalized_hypotonia [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Paroxysmal bursts of laughter (HP:0000749)	Poor_suck [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Paroxysmal bursts of laughter (HP:0000749)	Autism [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Seizures (HP:0001250)	Seizures [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Strabismus (HP:0000486)	Esotropia [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NDN (4692)	Widely spaced teeth (HP:0000687)	Carious_teeth [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NIPA1 (123606)	NIPA1 (123606)	Degeneration_of_the_lateral_corticospinal_tracts [HS]	MIM:600363
Angelman syndrome (MIM:105830)	NIPA1 (123606)	Muscular hypotonia (HP:0001252)	Spastic_gait [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NIPA1 (123606)	Muscular hypotonia (HP:0001252)	Spastic_paraplegia [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NIPA1 (123606)	Muscular hypotonia (HP:0001252)	Lower_limb_spasticity [HS]	MIM:176270
Angelman syndrome (MIM:105830)	NIPA1 (123606)	Seizures (HP:0001250)	Seizures [HS]	MIM:176270
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	variegated_coat_color [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	yellow_coat_color [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	abnormal_coat/hair_pigmentation [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	mottled_coat [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	delayed_hair_regeneration [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	abnormal_melanosome_morphology [MP]	PMID:20445456, PMID:10364509

Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	abnormal_hair_follicle_melanin_granule_morphology [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	abnormal_hair_follicle_melanin_granule_distribution [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	abnormal_melanogenesis [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	abnormal_hair_follicle_melanin_granule_shape [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	darkened_coat_color [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	premature_hair_loss [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	Red_hair [HS]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blond hair (HP:0002214)	diluted_coat_color [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blue irides (HP:0000635)	ocular_albinism [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blue irides (HP:0000635)	Albinism [HS]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blue irides (HP:0000635)	abnormal_melanosome_morphology [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blue irides (HP:0000635)	abnormal_melanogenesis [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blue irides (HP:0000635)	abnormal_choroid_melanin_granule_morphology [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blue irides (HP:0000635)	absent_eye_pigmentation [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Blue irides (HP:0000635)	abnormal_eye_pigmentation [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Broad-based_gait (HP:0002136)	abnormal_gait [MP]	-
Angelman syndrome (MIM:105830)	OCA2 (4948)	Hyperactivity (HP:0000752)	nervous [MP]	-
Angelman syndrome (MIM:105830)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	abnormal_ear_pigmentation [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	decreased_tail_pigmentation [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	Freckling [HS]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	abnormal_melanosome_morphology [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	abnormal_melanogenesis [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	abnormal_skin_pigmentation [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	Freckles_in_sun-exposed_areas [HS]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	reduced_eye_pigmentation [MP]	PMID:20445456, PMID:10364509
Angelman syndrome (MIM:105830)	OCA2 (4948)	Paroxysmal bursts of laughter (HP:0000749)	abnormal_pup_retrieval [MP]	-
Angelman syndrome (MIM:105830)	OCA2 (4948)	Paroxysmal bursts of laughter (HP:0000749)	abnormal_maternal_nurturing [MP]	-
Angelman syndrome (MIM:105830)	OCA2 (4948)	Progressive gait ataxia (HP:0007240)	abnormal_gait [MP]	-
Angelman syndrome (MIM:105830)	SNRPN (6638)	Absent speech development (HP:0001344)	Impaired_language_development [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Blond hair (HP:0002214)	Frontal_upsweep_of_hair [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Blond hair (HP:0002214)	Hypopigmentation_of_hair [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Blue irides (HP:000635)	Reduced_iris_pigmentation [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Flat occiput (HP:0005469)	Dolichocephaly [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Hyperactivity (HP:0000752)	Attention_deficit_hyperactivity_disorder [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Hypopigmentation of the skin (HP:0001010)	Hypopigmentation_of_the_skin [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Hypoplasia of the maxilla (HP:0000327)	Short_palm [HS]	-
Angelman syndrome (MIM:105830)	SNRPN (6638)	Microbrachycephaly (HP:0002258)	Dolichocephaly [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Motor delay (HP:0001270)	Motor_delay [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Muscular hypotonia (HP:0001252)	Generalized_hypotonia [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Paroxysmal bursts of laughter (HP:0000749)	Poor_suck [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Paroxysmal bursts of laughter (HP:0000749)	abnormal_suckling_behavior [MP]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Paroxysmal bursts of laughter (HP:0000749)	Autism [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Seizures (HP:0001250)	Seizures [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Strabismus (HP:0000486)	Esotropia [HS]	MIM:176275
Angelman syndrome (MIM:105830)	SNRPN (6638)	Widely spaced teeth (HP:0000687)	Carious_teeth [HS]	MIM:176275
Angelman syndrome (MIM:105830)	UBE3A (7337)	Broad-based_gait (HP:0002136)	abnormal_gait [MP]	PMID:12566516
Angelman syndrome (MIM:105830)	UBE3A (7337)	Cerebral_cortical_atrophy (HP:0002120)	abnormal_cerebral_cortex_morphology [MP]	PMID:8988171
Angelman syndrome (MIM:105830)	UBE3A (7337)	Progressive_gait_ataxia (HP:0007240)	abnormal_gait [MP]	PMID:12566516
Angelman syndrome (MIM:105830)	UBE3A (7337)	Progressive_gait_ataxia (HP:0007240)	small_cerebellum [MP]	PMID:12566516
Angelman syndrome (MIM:105830)	UBE3A (7337)	Seizures (HP:0001250)	tonic-clonic_seizures [MP]	PMID:10364509
Angelman syndrome (MIM:105830)	UBE3A (7337)	Seizures (HP:0001250)	audiogenic_seizures [MP]	PMID:10364509
Angelman syndrome (MIM:105830)	UBE3A (7337)	Seizures (HP:0001250)	absence_seizures [MP]	PMID:10364509
Angelman syndrome (MIM:105830)	UBE3A (7337)	Seizures (HP:0001250)	abnormal_spike_wave_discharge [MP]	PMID:10364509
Cri du Chat syndrome (MIM:123450)	CCT5 (22948)	High axial_irradius (HP:0001042)	distal_ulceration_and_osteomyelitis_leading_to_autoamputation [HS]	MIM:256840
Cri du Chat syndrome (MIM:123450)	CCT5 (22948)	Hypertonia (HP:0001276)	Spastic_gait [HS]	MIM:256840
Cri du Chat syndrome (MIM:123450)	CCT5 (22948)	Hypertonia (HP:0001276)	Lower_limb_spasticity [HS]	MIM:256840
Cri du Chat syndrome (MIM:123450)	CCT5 (22948)	Hypertonia (HP:0001276)	Spastic_paraplegia [HS]	MIM:256840
Cri du Chat syndrome (MIM:123450)	CCT5 (22948)	Microcephaly (HP:0000252)	abnormally_decreased_size_head [ZP]	MIM:256840
Cri du Chat syndrome (MIM:123450)	CCT5 (22948)	Short metacarpel (HP:0001049)	distal_ulceration_and_osteomyelitis_leading_to_autoamputation [HS]	MIM:256840
Cri du Chat syndrome (MIM:123450)	CCT5 (22948)	Short metatarsal bone (HP:00010743)	Abnormality_of_the_feet [HS]	MIM:256840
Cri du Chat syndrome (MIM:123450)	CCT5 (22948)	Single transverse palmar crease (HP:0000954)	distal_ulceration_and_osteomyelitis_leading_to_autoamputation [HS]	MIM:256840
Cri du Chat syndrome (MIM:123450)	IRX1 (79192)	Premature graying of hair (HP:0002216)	abnormally_increased_pigmentation_whole_organism [ZP]	MIM:256840
Cri du Chat syndrome (MIM:123450)	MTRR (4552)	Intellectual disability (HP:0001249)	Intellectual_disability_progressive [HS]	MIM:236270
Cri du Chat syndrome (MIM:123450)	MTRR (4552)	Neonatal hypotonia (HP:0001319)	Muscular_hypotonia [HS]	MIM:236270
Cri du Chat syndrome (MIM:123450)	NDUFS6 (4726)	Difficulty_walking (HP:0002355)	Exercise_intolerance [HS]	MIM:252010
Cri du Chat syndrome (MIM:123450)	NDUFS6 (4726)	Downslanted_palpebral_fissures (HP:0000494)	Ptosis [HS]	MIM:252010
Cri du Chat syndrome (MIM:123450)	NDUFS6 (4726)	Epicanthus (HP:0000286)	Ptosis [HS]	MIM:252010
Cri du Chat syndrome (MIM:123450)	NDUFS6 (4726)	Feeding_problems_in_infancy (HP:0008872)	Feeding_difficulties [HS]	MIM:252010
Cri du Chat syndrome (MIM:123450)	NDUFS6 (4726)	Hyperacusis (HP:0010780)	Sensorineural_hearing_impairment [HS]	MIM:252010
Cri du Chat syndrome (MIM:123450)	NDUFS6 (4726)	Hypertonia (HP:0010276)	Spasticity [HS]	MIM:252010
Cri du Chat syndrome (MIM:123450)	NDUFS6 (4726)	Microcephaly (HP:0000252)	Macrocephaly_progressive [HS]	MIM:252010
Cri du Chat syndrome (MIM:123450)	NDUFS6 (4726)	Neonatal hypotonia (HP:0001319)	Muscular_hypotonia [HS]	MIM:252010
Cri du Chat syndrome (MIM:123450)	NDUFS6 (4726)	Strabismus (HP:0000486)	Strabismus [HS]	MIM:252010
Cri du Chat syndrome (MIM:123450)	NKD2 (85409)	Broad_nasal_bridge (HP:0000431)	short_nasal_bone [MP]	-
Cri du Chat syndrome (MIM:123450)	NKD2 (85409)	Microretrognathia (HP:0000308)	decreased_cranium_height [MP]	-
Cri du Chat syndrome (MIM:123450)	NKD2 (85409)	Self-mutilation (HP:000742)	pup_cannibalization [MP]	-
Cri du Chat syndrome (MIM:123450)	SDHA (6389)	Anxiety (HP:0000739)	Emotional_ability [HS]	MIM:256000
Cri du Chat syndrome (MIM:123450)	SDHA (6389)	Difficulty_walking (HP:0002355)	Exercise_intolerance [HS]	MIM:256000
Cri du Chat syndrome (MIM:123450)	SDHA (6389)	Echolalia (HP:0010529)	Dysarthria [HS]	MIM:256000
Cri du Chat syndrome (MIM:123450)	SDHA (6389)	Hyperacusis (HP:0010780)	Bilateral_sensorineural_deafness [HS]	MIM:256000
Cri du Chat syndrome (MIM:123450)	SDHA (6389)	Hypertonia (HP:0001276)	Dystonia [HS]	MIM:256000, MIM:252011
Cri du Chat syndrome (MIM:123450)	SDHA (6389)	Hypertonia (HP:0001276)	Spasticity [HS]	MIM:256000, MIM:252011
Cri du Chat syndrome (MIM:123450)	SDHA (6389)	Intellectual_disability (HP:0001249)	Intellectual_disability [HS]	MIM:256000
Cri du Chat syndrome (MIM:123450)	SDHA (6389)	Neonatal_hypotonia (HP:0001319)	Dystonia [HS]	MIM:256000
Cri du Chat syndrome (MIM:123450)	SDHA (6389)	Neonatal_hypotonia (HP:0001319)	Neonatal_hypotonia [HS]	MIM:256000
Cri du Chat syndrome (MIM:123450)	SDHA (6389)	Premature graying of hair (HP:0002216)	Hypertrichosis [HS]	MIM:256000
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Hyperacusis (HP:0010780)	deafness [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Hyperacusis (HP:0010780)	decreased_brainstem_auditory_evoked_potential [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Hyperacusis (HP:0010780)	impaired_hearing [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Low-set_ears (HP:0000369)	absent Cochlear_outer_hair_cells [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Low-set_ears (HP:0000369)	cochlear_outer_hair_cell_degeneration [MP]	-

Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Low-set ears (HP:0000369)	organ_of_Corti_degeneration [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Low-set ears (HP:0000369)	absent_organ_of_Corti [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Low-set ears (HP:0000369)	abnormal_inner_ear_morphology [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Poorly formed pinnae (HP:0008562)	absent_cochlear_outer_hair_cells [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Poorly formed pinnae (HP:0008562)	cochlear_outer_hair_cell_degeneration [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Poorly formed pinnae (HP:0008562)	organ_of_Corti_degeneration [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Poorly formed pinnae (HP:0008562)	absent_organ_of_Corti [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC12A7 (10723)	Poorly formed pinnae (HP:0008562)	abnormal_inner_ear_morphology [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A19 (340024)	Anxiety (HP:0000739)	Emotional_lability [HS]	MIM:234500
Cri du Chat syndrome (MIM:123450)	SLC6A19 (340024)	Hypertonia (HP:0001276)	Hypertonia [HS]	MIM:234500
Cri du Chat syndrome (MIM:123450)	SLC6A19 (340024)	Intellectual_disability (HP:0001249)	Intellectual_disability [HS]	MIM:234500
Cri du Chat syndrome (MIM:123450)	SLC6A19 (340024)	Low birth weight (HP:0001518)	weight_loss [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A19 (340024)	Anxiety (HP:0000739)	abnormal_response_to_novel odor [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Anxiety (HP:0000739)	abnormal_response_to_novel_object [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Anxiety (HP:0000739)	increased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Anxiety (HP:0000739)	increased_thigmotaxis [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Anxiety (HP:0000739)	decreased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Autism (HP:0000717)	abnormal_response_to_novel odor [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Autism (HP:0000717)	abnormal_response_to_novel_object [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Autism (HP:0000717)	increased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Autism (HP:0000717)	decreased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Conspicuously happy disposition (HP:0100024)	abnormal_response_to_novel odor [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Conspicuously happy disposition (HP:0100024)	abnormal_response_to_novel_object [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Conspicuously happy disposition (HP:0100024)	increased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Conspicuously happy disposition (HP:0100024)	decreased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Difficulty walking (HP:0002355)	short_stride_length [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Echolalia (HP:0010529)	abnormal_response_to_novel odor [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Echolalia (HP:0010529)	abnormal_response_to_novel_object [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Echolalia (HP:0010529)	increased_stereotypic_behavior [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Echolalia (HP:0010529)	increased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Echolalia (HP:0010529)	decreased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Echolalia (HP:0010529)	induced_hyperactivity [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Hyperactivity (HP:0000752)	hyperactivity [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Hyperactivity (HP:0001276)	Limb_dystonia [HS]	MIM:613135
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Hyperactivity (HP:0001276)	Rigidity [HS]	MIM:613135
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Hyperactivity (HP:0001276)	hypertonia [MP]	MIM:613135
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Low birth weight (HP:0001518)	weight_loss [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Neonatal hypotonia (HP:0001319)	Muscular_hypotonia_of_the_trunk [HS]	MIM:613135
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Neonatal hypotonia (HP:0001319)	Limb_dystonia [HS]	MIM:613135
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Oppositional_defiant_disorder (HP:0010865)	abnormal_response_to_novel odor [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Oppositional_defiant_disorder (HP:0010865)	abnormal_response_to_novel_object [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Oppositional_defiant_disorder (HP:0010865)	increased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Oppositional_defiant_disorder (HP:0010865)	decreased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Overfriendliness (HP:0100025)	abnormal_response_to_novel odor [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Overfriendliness (HP:0100025)	abnormal_response_to_novel_object [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Overfriendliness (HP:0100025)	increased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Overfriendliness (HP:0100025)	decreased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Self-mutilation (HP:000742)	abnormal_maternal_nurturing [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Self-mutilation (HP:000742)	abnormal_response_to_novel odor [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Self-mutilation (HP:000742)	abnormal_response_to_novel_object [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Self-mutilation (HP:000742)	increased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Self-mutilation (HP:000742)	decreased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Self-mutilation (HP:000742)	increased_exploration_in_new_environment [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Short metatarsal bone (HP:0010743)	decreased_length_of_long_bones [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC6A3 (6531)	Stereotyped_repetitive behaviour (HP:0000733)	increased_stereotypic_behavior [MP]	-
Cri du Chat syndrome (MIM:123450)	SLC9A3 (6550)	Gastroesophageal reflux (HP:0002020)	esophageal_epithelium_hyperplasia [MP]	-
Cri du Chat syndrome (MIM:123450)	TERT (7015)	Broad nasal bridge (HP:0000431)	impaired_olfaction [MP]	-
Cri du Chat syndrome (MIM:123450)	TERT (7015)	Microretrognathia (HP:0000308)	decreased_nb_number [MP]	-
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Colon cancer (HP:0003003)	large_intestinal_inflammation [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Colon cancer (HP:0003003)	rectal_hemorrhage [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Colon cancer (HP:0003003)	Desmoid_tumors [HS]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Colon cancer (HP:0003003)	Colon_cancer [HS]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Colon cancer (HP:0003003)	Hepatocellular_carcinoma [HS, MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Colon cancer (HP:0003003)	rectal_prolapse [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Colon cancer (HP:0003003)	Hereditary_nonpolyposis_colorectal_carcinoma [HS]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Colon cancer (HP:0003003)	abnormal_large_intestine_crypts_of_Lieberkuhn_morphology [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Colon cancer (HP:0003003)	abnormal_colon_morphology [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple adenomatous colon polyps (HP:0005227)	colon_polyps [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple adenomatous colon polyps (HP:0005227)	gastric_polyps [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple adenomatous colon polyps (HP:0005227)	large_intestinal_inflammation [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple adenomatous colon polyps (HP:0005227)	rectal_hemorrhage [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple adenomatous colon polyps (HP:0005227)	Desmoid_tumors [HS]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple adenomatous colon polyps (HP:0005227)	Hepatocellular_carcinoma [HS, MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple adenomatous colon polyps (HP:0005227)	rectal_prolapse [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple adenomatous colon polyps (HP:0005227)	Hereditary_nonpolyposis_colorectal_carcinoma [HS]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple adenomatous colon polyps (HP:0005227)	abnormal_large_intestine_crypts_of_Lieberkuhn_morphology [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple adenomatous colon polyps (HP:0005227)	abnormal_colon_morphology [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple adenomatous colon polyps (HP:0005227)	intestine_polyps [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	multiple duodenal polyps (HP:0004783)	colon_polyps [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	multiple duodenal polyps (HP:0004783)	gastric_polyps [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	multiple duodenal polyps (HP:0004783)	Desmoid_tumors [HS]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	multiple duodenal polyps (HP:0004783)	Hepatocellular_carcinoma [HS, MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	multiple duodenal polyps (HP:0004783)	intestine_polyps [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple gastric polyps (HP:0004394)	small_stomach [MP]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple gastric polyps (HP:0004394)	Desmoid_tumors [HS]	PMID:1651174
Familial Adenomatous Polyposis (MIM:175100)	APC (324)	Multiple gastric polyps (HP:0004394)	Hepatocellular_carcinoma [HS, MP]	PMID:1651174

Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Abnormality of the carpal bones (HP:0001191)	Abnormality_of_the_hand [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Fibular hypoplasia (HP:0003038)	Micrognathia [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Fibular hypoplasia (HP:0003038)	Short_femoral_neck [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Fibular hypoplasia (HP:0003038)	Rudimentary_fibula [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Hypoplasia of the radius (HP:0002984)	Hypoplasia_of_the_radius [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Hypoplasia of the radius (HP:0002984)	Micrognathia [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Hypoplasia of the ulna (HP:0003022)	Hypoplasia_of_the_ulna [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Hypoplasia of the ulna (HP:0003022)	Micrognathia [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Hypoplastic tibia (HP:0005736)	Shortening_of_the_tibia [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Hypoplastic tibia (HP:0005736)	Micrognathia [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Hypoplastic tibia (HP:0005736)	Short_femoral_neck [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Madelung deformity (HP:0003067)	Madelung_deformity [HS]	PMID:11889216
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Mesomelia (HP:0003027)	Micrognathia [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Mesomelia (HP:0003027)	Mesomelia [HS]	PMID:14557470
Leri-Weill dyschondroostosis (MIM:127300)	SHOX (6473)	Radial bowing (HP:0002986)	Radial_bowing [HS]	PMID:14557470
Leri-Weill dyschondroostosis	SHOX (6473)	Short stature, disproportionate short-limbed	Short_stature,_mesomelic [HS]	PMID:10634394
Xq28 (MECP2) duplication (DECIPHER:45)	ABCD1 (215)	Spasticity, progressive (HP:0002191)	Spastic_paraplegia [HS]	MIM:300100
Xq28 (MECP2) duplication (DECIPHER:45)	AVPR2 (554)	Drooling (HP:0002307)	Vomiting [HS]	MIM:304800
Xq28 (MECP2) duplication (DECIPHER:45)	AVPR2 (554)	Dysphagia (HP:0002015)	Polydipsia [HS, MP]	MIM:304800
Xq28 (MECP2) duplication (DECIPHER:45)	AVPR2 (554)	Dysphagia (HP:0002015)	Feeding_difficulties [HS]	MIM:304800
Xq28 (MECP2) duplication (DECIPHER:45)	AVPR2 (554)	Intellectual disability, severe (HP:0010864)	Intellectual_disability [HS]	MIM:304800
Xq28 (MECP2) duplication (DECIPHER:45)	AVPR2 (554)	Seizures (HP:0001250)	Seizures [HS]	MIM:300539
Xq28 (MECP2) duplication (DECIPHER:45)	BGN (633)	Inability to walk (HP:0002540)	abnormal_gait [MP]	PMID:12975603
Xq28 (MECP2) duplication (DECIPHER:45)	L1CAM (3897)	Absent speech development (HP:0001344)	Aphasia [HS]	MIM:303350
Xq28 (MECP2) duplication (DECIPHER:45)	L1CAM (3897)	Inability to walk (HP:0002540)	Shuffling_gait [HS]	MIM:303350
Xq28 (MECP2) duplication (DECIPHER:45)	L1CAM (3897)	Intellectual disability, severe (HP:0010864)	Intellectual_disability [HS]	MIM:303350
Xq28 (MECP2) duplication (DECIPHER:45)	L1CAM (3897)	Seizures (HP:0001250)	Seizures [HS]	MIM:303350
Xq28 (MECP2) duplication (DECIPHER:45)	L1CAM (3897)	Spasticity, progressive (HP:0002191)	Lower_limb_spasticity [HS]	MIM:303350
Xq28 (MECP2) duplication (DECIPHER:45)	L1CAM (3897)	Spasticity, progressive (HP:0002191)	Spastic_paraplegia [HS]	MIM:303350
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Absent speech development (HP:0001344)	Lack_of_language_development [HS]	MIM:303350, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Absent speech development (HP:0001344)	Absent_speech_development [HS]	MIM:303350, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Absent speech development (HP:0001344)	Speech_delay [HS]	MIM:303350, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Drooling (HP:0002307)	Drooling [HS]	MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Dysphagia (HP:0002015)	abnormal_orbitofrontal_cortex_morphology [MP]	MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Dysphagia (HP:0002015)	Dysphagia [HS]	MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Dysphagia (HP:0002015)	Short_neck [HS]	MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Gastroesophageal reflux (HP:0002020)	abnormal_orbitofrontal_cortex_morphology [MP]	MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Gastroesophageal reflux (HP:0002020)	Gastroesophageal_reflux [HS]	MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Gastroesophageal reflux (HP:0002020)	Short_neck [HS]	MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Inability to walk (HP:0002540)	short_stride_length [MP]	PMID:15241799, MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Inability to walk (HP:0002540)	abnormal_gait [MP]	PMID:15241799, MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Inability to walk (HP:0002540)	Progressive_gait_ataxia [HS]	PMID:15241799, MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Inability to walk (HP:0002540)	Shuffling_gait [HS]	PMID:15241799, MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Inability to walk (HP:0002540)	Broad-based_gait [HS]	PMID:15241799, MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Inability to walk (HP:0002540)	Gait_apraxia [HS]	PMID:15241799, MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Intellectual disability, severe (HP:0010864)	Intellectual_disability,_profound [HS]	MIM:312750, MIM:300673, MIM:300055
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Intellectual disability, severe (HP:0010864)	Intellectual_disability,_progressive [HS]	MIM:312750, MIM:300673, MIM:300055
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Intellectual disability, severe (HP:0010864)	Intellectual_disability,_mild [HS]	MIM:312750, MIM:300673, MIM:300055
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Narrow mouth (HP:0000160)	Narrow_mouth [HS]	MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Neonatal hypotonia (HP:0001319)	Facial_hypotonia [HS]	MIM:312750, MIM:300673, MIM:303350, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Neonatal hypotonia (HP:0001319)	Dystonia [HS]	MIM:312750, MIM:300673, MIM:303350, MIM:300260

Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Neonatal hypotonia (HP:0001319)	Muscular_hypotonia_of_the_trunk [HS]	MIM:312750, MIM:300673, MIM:303350, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Neonatal hypotonia (HP:0001319)	Infantile_muscular_hypotonia [HS]	MIM:312750, MIM:300673, MIM:303350, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Recurrent respiratory infections (HP:0002205)	Recurrent_respiratory_infections [HS]	MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Seizures (HP:0001250)	Myoclonus [HS]	PMID:15241799, MIM:312750
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Seizures (HP:0001250)	sporadic_seizures [MP]	PMID:15241799, MIM:312750
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Seizures (HP:0001250)	Seizures [HS, MP]	PMID:15241799, MIM:312750
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Spasmodic, progressive (HP:0002191)	Spasmodic,_progressive [HS]	MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Spasmodic, progressive (HP:0002191)	Dystonia [HS]	MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Spasmodic, progressive (HP:0002191)	Spastic_gait [HS]	MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	MECP2 (4204)	Spasmodic, progressive (HP:0002191)	Rigidity [HS]	MIM:312750, MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	SLC6A8 (6535)	Absent speech development (HP:0001344)	Speech_and_language_delay,_severe [HS]	MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	SLC6A8 (6535)	Drooling (HP:0002307)	Vomiting [HS]	MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	SLC6A8 (6535)	Dysphagia (HP:0002015)	Feeding_difficulties [HS]	MIM:300673
Xq28 (MECP2) duplication (DECIPHER:45)	SLC6A8 (6535)	Neonatal hypotonia (HP:0001319)	Neonatal_hypotonia [HS]	MIM:300260
Xq28 (MECP2) duplication (DECIPHER:45)	SLC6A8 (6535)	Seizures (HP:0001250)	Seizures [HS]	MIM:312750
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Low-set ears (HP:0000369)	abnormal_vestibule_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Low-set ears (HP:0000369)	abnormal_ear_development [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Low-set ears (HP:0000369)	absent_otooliths [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Low-set ears (HP:0000369)	decreased_otoolith_number [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Low-set ears (HP:0000369)	abnormal_utricle_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Low-set ears (HP:0000369)	enlarged_otooliths [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Low-set ears (HP:0000369)	abnormal_vestibular_saccule_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Posteriorly rotated ears (HP:0000358)	abnormal_vestibule_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Posteriorly rotated ears (HP:0000358)	abnormal_ear_development [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Posteriorly rotated ears (HP:0000358)	absent_otooliths [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Posteriorly rotated ears (HP:0000358)	decreased_otoolith_number [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Posteriorly rotated ears (HP:0000358)	abnormal_utricle_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Posteriorly rotated ears (HP:0000358)	enlarged_otooliths [MP]	-
Miller-Dieker syndrome (MIM:247200)	ABR (29)	Posteriorly rotated ears (HP:0000358)	abnormal_vestibular_saccule_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	CRK (1398)	Cleft palate (HP:0000175)	palatal_shelves_fail_to_meet_at_midline [MP]	PMID:12905154
Miller-Dieker syndrome (MIM:247200)	CRK (1398)	Cleft palate (HP:0000175)	cleft_secondary_palate [MP]	PMID:12905154
Miller-Dieker syndrome (MIM:247200)	CRK (1398)	Hypoplasia of the corpus callosum (HP:0002079)	abnormal_hippocampus_morphology [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	CRK (1398)	Hypoplasia of the corpus callosum (HP:0002079)	abnormal_dentate_gyrus_morphology [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	CRK (1398)	Hypoplasia of the corpus callosum (HP:0002079)	abnormal_hippocampus_pyramidal_cell_layer [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	CRK (1398)	Lissencephaly (HP:0001339)	abnormal_cerebral_cortex_pyramidal_cell_morphology [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	CRK (1398)	Lissencephaly (HP:0001339)	abnormal_cerebral_cortex_morphology [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	CRK (1398)	Nasal hypoplasia (HP:0003196)	abnormal_nose_morphology [MP]	PMID:12905154
Miller-Dieker syndrome (MIM:247200)	CRK (1398)	Nasal hypoplasia (HP:0003196)	abnormal_nasal_mucosa_morphology [MP]	PMID:12905154
Miller-Dieker syndrome (MIM:247200)	CRK (1398)	Pachygyria (HP:0001302)	abnormal_cerebral_cortex_pyramidal_cell_morphology [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	CRK (1398)	Pachygyria (HP:0001302)	abnormal_cerebral_cortex_morphology [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	DPH1 (1801)	Cleft palate (HP:0000175)	cleft_palate [MP]	-
Miller-Dieker syndrome (MIM:247200)	DPH1 (1801)	Clindactyly of the 5th finger (HP:0004209)	polydactyly [MP]	-
Miller-Dieker syndrome (MIM:247200)	DPH1 (1801)	Intrauterine growth restriction (HP:0001511)	embryonic_growth_retardation [MP]	PMID:12905154
Miller-Dieker syndrome (MIM:247200)	HIC1 (3090)	Cleft palate (HP:0000175)	abnormal_palate_morphology [MP]	PMID:10655551
Miller-Dieker syndrome (MIM:247200)	HIC1 (3090)	Inguinal hernia (HP:0000023)	abnormal_ventral_body_wall_morphology [MP]	PMID:10655551
Miller-Dieker syndrome (MIM:247200)	HIC1 (3090)	Low-set ears (HP:0000369)	lowered_ear_position [MP]	PMID:10655551
Miller-Dieker syndrome (MIM:247200)	HIC1 (3090)	Low-set ears (HP:0000369)	small_ears [MP]	PMID:10655551
Miller-Dieker syndrome (MIM:247200)	HIC1 (3090)	Nasal hypoplasia (HP:0003196)	short_snout [MP]	PMID:10655551
Miller-Dieker syndrome (MIM:247200)	HIC1 (3090)	Omphalocele (HP:0001539)	abnormal_ventral_body_wall_morphology [MP]	PMID:10655551
Miller-Dieker syndrome (MIM:247200)	HIC1 (3090)	Posteriorly rotated ears (HP:0000358)	small_ears [MP]	PMID:10655551
Miller-Dieker syndrome (MIM:247200)	MNT (4335)	Cleft palate (HP:0000175)	cleft_palate [MP]	PMID:12905154
Miller-Dieker syndrome (MIM:247200)	MNT (4335)	Failure of palatal shelf elevation (HP:0000175)	failure_of_palatal_shelf_elevation [MP]	PMID:12905154
Miller-Dieker syndrome (MIM:247200)	MNT (4335)	Micrognathia (HP:0000347)	mandible_hypoplasia [MP]	PMID:12905154
Miller-Dieker syndrome (MIM:247200)	MNT (4335)	Micrognathia (HP:0000347)	small_cranium [MP]	PMID:12905154
Miller-Dieker syndrome (MIM:247200)	MYO1C (4641)	Heterotopia (HP:0002282)	abnormal Vestibular hair cell physiology [MP]	-
Miller-Dieker syndrome (MIM:247200)	MYO1C (4641)	Lissencephaly (HP:0001339)	abnormal Vestibular hair cell physiology [MP]	-
Miller-Dieker syndrome (MIM:247200)	MYO1C (4641)	Pachygyria (HP:0001302)	abnormal Vestibular hair cell physiology [MP]	-
Miller-Dieker syndrome (MIM:247200)	NXN (64359)	Joint contractures involving the joints of the hand (HP:0009473)	abnormal_sternum_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	NXN (64359)	Joint contractures involving the joints of the hand (HP:0009473)	split_xiphoid_process [MP]	-
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Cryptorchidism (HP:0000028)	abnormal_seminiferous_epithelium_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Cryptorchidism (HP:0000028)	abnormal_acrosome_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Cryptorchidism (HP:0000028)	abnormal_seminiferous_tubule_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Cryptorchidism (HP:0000028)	abnormal_testis_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Cryptorchidism (HP:0000028)	testis_hypoplasia [MP]	-
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Cryptorchidism (HP:0000028)	decreased testis_weight [MP]	-
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Heterotopia (HP:0002282)	abnormal_neuronal_migration [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Heterotopia (HP:0002282)	increased_neuron_apoptosis [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Heterotopia (HP:0002282)	Heterotopia [HS]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Hypoplasia of the corpus callosum (HP:0002079)	abnormal_hippocampus_morphology [MP]	MIM:607432
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Hypoplasia of the corpus callosum (HP:0002079)	abnormal_dentate_gyrus_morphology [MP]	MIM:607432
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Hypoplasia of the corpus callosum (HP:0002079)	abnormal_hippocampus_layer_morphology [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Hypoplasia of the corpus callosum (HP:0002079)	abnormal_hippocampus_pyramidal_cell_layer [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Hypoplasia of the corpus callosum (HP:0002079)	Abnormality_of_the_cerebral_white_matter [HS]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Hypoplasia of the corpus callosum (HP:0002079)	abnormal_hippocampus_pyramidal_cell_morphology [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Hypoplasia of the corpus callosum (HP:0002079)	absent_hippocampus_stratum_orientalis [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Intellectual disability (HP:0001249)	Intellectual_disability [HS]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Lissencephaly (HP:0001339)	abnormal Stratification_in_cerebral_cortex [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Lissencephaly (HP:0001339)	abnormal_cerebral_cortex_morphology [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Lissencephaly (HP:0001339)	abnormal_neocortex_morphology [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Lissencephaly (HP:0001339)	abnormal_neuronal_migration [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Lissencephaly (HP:0001339)	increased_neuron_apoptosis [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Lissencephaly (HP:0001339)	Lissencephaly [HS]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Microcephaly (HP:0000252)	Microcephaly,_postnatal [HS]	MIM:607432
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Pachygyria (HP:0001302)	abnormal Stratification_in_cerebral_cortex [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Pachygyria (HP:0001302)	abnormal_cerebral_cortex_morphology [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Pachygyria (HP:0001302)	abnormal_neocortex_morphology [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Pachygyria (HP:0001302)	abnormal_neuronal_migration [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Pachygyria (HP:0001302)	increased_neuron_apoptosis [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Pachygyria (HP:0001302)	Pachygyria [HS]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Pachygyria (HP:0001302)	increased_neuron_apoptosis [MP]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Progressive spastic paraparesia (HP:0007020)	Muscular_hypotonia_of_the_trunk [HS]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Progressive spastic paraparesia (HP:0007020)	Spastic_tetraparesis [HS]	PMID:9063735
Miller-Dieker syndrome (MIM:247200)	PAFAH1B1 (5048)	Seizures (HP:0001250)	Seizures [HS]	PMID:9063735

Miller-Dieker syndrome (MIM:247200)	PITPNA (5306)	Broad nasal bridge (HP:0000431)	abnormally_present_in_fewer_numbers_in_organism_spinal_cord sensory_neuron [ZP]	-
Miller-Dieker syndrome (MIM:247200)	PITPNA (5306)	Nasal hypoplasia (HP:0003196)	abnormally_present_in_fewer_numbers_in_organism_spinal_cord_sensory_neuron [ZP]	-
Miller-Dieker syndrome (MIM:247200)	PITPNA (5306)	Seizures (HP:0001250)	seizures [MP]	-
Miller-Dieker syndrome (MIM:247200)	RPA1 (6117)	Midline brain calcifications (HP:0007045)	abnormally_necrotic_brain [ZP]	-
Miller-Dieker syndrome (MIM:247200)	SERPINF1 (5176)	Cryptorchidism (HP:0000028)	abnormal_prostate_gland_morphology [MP]	-
Miller-Dieker syndrome (MIM:247200)	SERPINF1 (5176)	Cryptorchidism (HP:0000028)	prostate_gland_epithelial_hyperplasia [MP]	-
Miller-Dieker syndrome (MIM:247200)	SERPINF2 (5345)	Inguinal hernia (HP:0000023)	Abnormality_of_the_thorax [HS]	PMID:82839
Miller-Dieker syndrome (MIM:247200)	SERPINF2 (5345)	Omphalocele (HP:0001539)	Abnormality_of_the_thorax [HS]	PMID:82839
Miller-Dieker syndrome (MIM:247200)	SRR (63826)	Heterotopia (HP:0002282)	decreased_susceptibility_to_neuronal_excitotoxicity [MP]	-
Miller-Dieker syndrome (MIM:247200)	SRR (63826)	Lissencephaly (HP:0001339)	decreased_susceptibility_to_neuronal_excitotoxicity [MP]	-
Miller-Dieker syndrome (MIM:247200)	SRR (63826)	Pachygyria (HP:0001302)	decreased_susceptibility_to_neuronal_excitotoxicity [MP]	-
Miller-Dieker syndrome (MIM:247200)	VPS53 (55275)	Intrauterine growth restriction (HP:0001511)	embryonic_growth_retardation [MP]	-
Miller-Dieker syndrome (MIM:247200)	YWHAE (7531)	Heterotopia (HP:0002282)	abnormal_neuronal_migration [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	YWHAE (7531)	Hypoplasia_of_the_cortex callosum (HP:0002079)	abnormal_hippocampus_morphology [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	YWHAE (7531)	Hypoplasia_of_the_cortex callosum (HP:0002079)	abnormal_hippocampus_pyramidal_cell_layer [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	YWHAE (7531)	Lissencephaly (HP:0001339)	abnormal_neuronal_migration [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	YWHAE (7531)	Lissencephaly (HP:0001339)	abnormal_cerebral_cortex_morphology [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	YWHAE (7531)	Pachygyria (HP:0001302)	abnormal_neuronal_migration [MP]	PMID:12621583
Miller-Dieker syndrome (MIM:247200)	YWHAE (7531)	Pachygyria (HP:0001302)	abnormal_cerebral_cortex_morphology [MP]	PMID:12621583
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Lisch nodules (HP:0009737)	increased_gland_tumor_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Lisch nodules (HP:0009737)	T_cell_derived_lymphoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Lisch nodules (HP:0009737)	increased_ovary_tumor_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Lisch nodules (HP:0009737)	uterus_tumor [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Lisch nodules (HP:0009737)	increased_lymphoma_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	increased_gland_tumor_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	liver_adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	T_cell_derived_lymphoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	increased_spindle_cell_carcinoma_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	hemangiosarcoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	increased_ovary_tumor_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	uterus_tumor [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	lung_carcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	increased_lymphoma_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	lung_adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Neurofibrosarcoma (HP:0100697)	sarcoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Plexiform neurofibroma (HP:0009732)	liver_adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Plexiform neurofibroma (HP:0009732)	T_cell_derived_lymphoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Plexiform neurofibroma (HP:0009732)	increased_spindle_cell_carcinoma_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Plexiform neurofibroma (HP:0009732)	hemangiosarcoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Plexiform neurofibroma (HP:0009732)	lung_carcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Plexiform neurofibroma (HP:0009732)	adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Plexiform neurofibroma (HP:0009732)	increased_lymphoma_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Plexiform neurofibroma (HP:0009732)	lung_adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Plexiform neurofibroma (HP:0009732)	sarcoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Spinal neurofibromas (HP:0009735)	liver_adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Spinal neurofibromas (HP:0009735)	T_cell_derived_lymphoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Spinal neurofibromas (HP:0009735)	increased_spindle_cell_carcinoma_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Spinal neurofibromas (HP:0009735)	hemangiosarcoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Spinal neurofibromas (HP:0009735)	lung_carcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Spinal neurofibromas (HP:0009735)	adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Spinal neurofibromas (HP:0009735)	increased_lymphoma_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Spinal neurofibromas (HP:0009735)	lung_adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Subcutaneous neurofibromas (HP:0100698)	sarcoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Subcutaneous neurofibromas (HP:0100698)	liver_adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Subcutaneous neurofibromas (HP:0100698)	T_cell_derived_lymphoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Subcutaneous neurofibromas (HP:0100698)	increased_spindle_cell_carcinoma_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Subcutaneous neurofibromas (HP:0100698)	hemangiosarcoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Subcutaneous neurofibromas (HP:0100698)	lung_carcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Subcutaneous neurofibromas (HP:0100698)	adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Subcutaneous neurofibromas (HP:0100698)	increased_lymphoma_incidence [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Subcutaneous neurofibromas (HP:0100698)	lung_adenocarcinoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	ATAD5 (79915)	Subcutaneous neurofibromas (HP:0100698)	sarcoma [MP]	PMID:21901109
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Axillary freckling (HP:0000997)	reduced_eye_pigmentation [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Axillary freckling (HP:0000997)	Axillary_freckling [HS]	PMID:14729829

NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Broad neck (HP:0000475)	Webbed_neck [HS]	MIM:601321, MIM:162210
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Broad neck (HP:0000475)	Parathyroid_adenoma [HS]	PMID:8116612
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Cafe-au-lait spots (HP:0000957)	Multiple_cafe-au-lait_spots [HS]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Cafe-au-lait spots (HP:0000957)	reduced_eye_pigmentation [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Coarse facial features (HP:0000280)	Malar_hypoplasia [HS]	MIM:162200, MIM:193520
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Hypertelorism (HP:0000316)	Hypertelorism [HS]	PMID:8116612
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Learning disability (HP:0001328)	Learning_disability [HS]	PMID:8116612
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Lisch nodules (HP:0009737)	Lisch_nodules [HS]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Lisch nodules (HP:0009737)	increased_hepatoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Lisch nodules (HP:0009737)	leukemia [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Lisch nodules (HP:0009737)	increased_fibrohistiocytoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Lisch nodules (HP:0009737)	increased_lymphoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Lisch nodules (HP:0009737)	increased_adrenal_gland_tumor_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Macrocephaly (HP:0000256)	Relative_macrocephaly [HS]	MIM:162200
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Macrocephaly (HP:0000256)	megacephaly [MP]	MIM:162210
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	brain_tumor [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	increased_neurofibroma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	increased_gangliosarcoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	increased_fibrosarcoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	increased_ganglioneuroma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	increased_neurofibrosarcoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	increased_hepatoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	increased_astrocytoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	increased_glioma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	leiomyosarcoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	sarcoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	leukemia [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	increased_globioblastoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	malignant_triton_tumors [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	carcinoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	Rhabdomyosarcoma [HS, MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	increased_lymphoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	lung_adenocarcinoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Neurofibrosarcoma (HP:0100697)	increased_adrenal_gland_tumor_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Optic glioma (HP:0009734)	abnormal_astrocyte_morphology [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Optic glioma (HP:0009734)	Optic_glioma [HS]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Optic glioma (HP:0009734)	Spinal_cord_tumor [HS]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Optic glioma (HP:0009734)	astrocytosis [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Optic glioma (HP:0009734)	neuroblastoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Optic glioma (HP:0009734)	Astrocytoma [HS]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Optic glioma (HP:0009734)	increased_oligodendrocyte_number [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Optic glioma (HP:0009734)	Meningioma [HS]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Pectus excavatum (HP:0000767)	enlarged_chest [MP]	MIM:162200
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Pes cavus (HP:0001761)	Tibial_pseudoarthrosis [HS]	MIM:162200
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Pes cavus (HP:0001761)	Lower_limb_muscle_weakness [HS]	MIM:601321
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Pes cavus (HP:0001761)	Genu_valgum [HS]	MIM:601321
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Pes cavus (HP:0001761)	Paraparesis [HS]	PMID:8116612
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Plexiform neurofibroma (HP:0009732)	Plexiform_neurofibroma [HS]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Plexiform neurofibroma (HP:0009732)	Pheochromocytoma [HS, MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Plexiform neurofibroma (HP:0009732)	leiomyosarcoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Plexiform neurofibroma (HP:0009732)	sarcoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Plexiform neurofibroma (HP:0009732)	leukemia [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Plexiform neurofibroma (HP:0009732)	malignant_triton_tumors [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Plexiform neurofibroma (HP:0009732)	carcinoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Plexiform neurofibroma (HP:0009732)	Rhabdomyosarcoma [HS, MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Plexiform neurofibroma (HP:0009732)	increased_lymphoma_incidence [MP]	PMID:14729829

NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Plexiform neurofibroma (HP:0009732)	lung_adenocarcinoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Scoliosis (HP:0002650)	Scoliosis [HS]	MIM:162200
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Spinal neurofibromas (HP:0009735)	Pheochromocytoma [HS, MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Spinal neurofibromas (HP:0009735)	leiomyosarcoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Spinal neurofibromas (HP:0009735)	sarcoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Spinal neurofibromas (HP:0009735)	leukemia [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Spinal neurofibromas (HP:0009735)	Symmetric_spinal_nerve_root_neurofibromas [HS]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Spinal neurofibromas (HP:0009735)	malignant_triton_tumors [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Spinal neurofibromas (HP:0009735)	carcinoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Spinal neurofibromas (HP:0009735)	Rhabdomyosarcoma [HS, MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Spinal neurofibromas (HP:0009735)	increased_lymphoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Spinal neurofibromas (HP:0009735)	lung_adenocarcinoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Subcutaneous neurofibromas (HP:0100698)	Pheochromocytoma [HS, MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Subcutaneous neurofibromas (HP:0100698)	leiomyosarcoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Subcutaneous neurofibromas (HP:0100698)	sarcoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Subcutaneous neurofibromas (HP:0100698)	Rhabdomyosarcoma [HS, MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Subcutaneous neurofibromas (HP:0100698)	leukemia [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Subcutaneous neurofibromas (HP:0100698)	malignant_triton_tumors [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Subcutaneous neurofibromas (HP:0100698)	carcinoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Subcutaneous neurofibromas (HP:0100698)	Rhabdomyosarcoma [HS, MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Subcutaneous neurofibromas (HP:0100698)	increased_lymphoma_incidence [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Subcutaneous neurofibromas (HP:0100698)	lung_adenocarcinoma [MP]	PMID:14729829
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Tall stature (HP:0000098)	Overgrowth [HS]	MIM:193520, MIM:162200
NF1-microdeletion syndrome (MIM:613675)	NF1 (4763)	Tall stature (HP:0000098)	Short_stature [HS]	MIM:193520, MIM:162200
NF1-microdeletion syndrome (MIM:613675)	RAB11FIP4 (84440)	Macrocephaly (HP:0000256)	abnormally_decreased_size_head [ZP]	-
Pelizaeus-Merzbacher disease (MIM:312080)	GLRA4 (441509)	Hyporeflexia (HP:0001265)	abnormally_disrupted_cellular_response_to_mechanical_stimulus [ZP]	-
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Abnormal myelination (HP:0002520)	abnormally_myelination [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Ataxia (HP:0001251)	ataxia [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Ataxia (HP:0001251)	Dysmetria [HS]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Ataxia (HP:0001251)	abnormal_cerebellar_granule_layer [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Ataxia (HP:0001251)	Spinocerebellar_tract_degeneration [HS]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Ataxia (HP:0001251)	abnormal_cerebellum_white_matter_morphology [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Ataxia (HP:0001251)	abnormal_cerebellum_morphology [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Dysarthria (HP:0001260)	Dysarthria [HS]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Hyporeflexia (HP:0001265)	Hyperreflexia [HS]	MIM:312920
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Nystagmus (HP:0000639)	abnormal_startle_reflex [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Nystagmus (HP:0000639)	Nystagmus [HS]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Nystagmus (HP:0000639)	tremors [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Optic atrophy (HP:0000648)	abnormal_optic_nerve_morphology [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Optic atrophy (HP:0000648)	Optic_atrophy [HS]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Progressive spastic quadriplegia (HP:0002478)	Spastic_gait [HS]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Progressive spastic quadriplegia (HP:0002478)	Lower_limb_spasticity [HS]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Progressive spastic quadriplegia (HP:0002478)	Spastic_paraplegia [HS]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Pyramidal signs (HP:0007256)	Degeneration_of_the_lateral_corticospinal_tracts [HS]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Pyramidal signs (HP:0007256)	Babinski_sign [HS]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Reduction of oligodendroglia (HP:0100709)	abnormal_microglial_cell_morphology [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Reduction of oligodendroglia (HP:0100709)	abnormal_CNS_gliai_cell_morphology [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Reduction of oligodendroglia (HP:0100709)	astrocytosis [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Reduction of oligodendroglia (HP:0100709)	increased_oligodendrocyte_number [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Reduction of oligodendroglia (HP:0100709)	abnormal_astrocyte_morphology [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Reduction of oligodendroglia (HP:0100709)	gliosis [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Reduction of oligodendroglia (HP:0100709)	decreased_oligodendrocyte_number [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Reduction of oligodendroglia (HP:0100709)	abnormal_oligodendrocyte_morphology [MP]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Sudanophilic leukodystrophy (HP:0003269)	Sudanophilic_leukodystrophy [HS]	PMID:7915877
Pelizaeus-Merzbacher disease (MIM:312080)	PLP1 (5354)	Sudanophilic leukodystrophy (HP:0003269)	abnormal_nerve_conduction [MP]	PMID:7915877
Phelan-McDermid syndrome (MIM:606232)	ARSA (410)	Aggressive behavior (HP:0000718)	Emotional_lability [HS]	MIM:250100

Phelan-Mcdermid syndrome (MIM:606232)	ARSA (410)	Autism (HP:0000717)	Emotional_lability [HS]	MIM:250100
Phelan-Mcdermid syndrome (MIM:606232)	ARSA (410)	Broad-based gait (HP:0002136)	short_stride_length [MP]	MIM:250100
Phelan-Mcdermid syndrome (MIM:606232)	ARSA (410)	Broad-based gait (HP:0002136)	Gait_disturbance [HS, MP]	MIM:250100
Phelan-Mcdermid syndrome (MIM:606232)	ARSA (410)	Hearing impairment (HP:0000365)	deafness [MP]	-
Phelan-Mcdermid syndrome (MIM:606232)	ARSA (410)	Hearing impairment (HP:0000365)	absent_brainstem_auditory_evoked_potential [MP]	-
Phelan-Mcdermid syndrome (MIM:606232)	ARSA (410)	Neonatal hypotonia (HP:0001319)	Muscular_hypotonia [HS]	MIM:250100
Phelan-Mcdermid syndrome (MIM:606232)	ARSA (410)	Neonatal hypotonia (HP:0001319)	Spastic_tetraplegia [HS]	MIM:250100
Phelan-Mcdermid syndrome (MIM:606232)	ARSA (410)	Neonatal hypotonia (HP:0001319)	Dystonia [HS]	MIM:250100
Phelan-Mcdermid syndrome (MIM:606232)	ARSA (410)	Seizures (HP:0001250)	Seizures [HS]	MIM:250100
Phelan-Mcdermid syndrome (MIM:606232)	ARSA (410)	Unsteady gait (HP:0002317)	short_stride_length [MP]	MIM:250100
Phelan-Mcdermid syndrome (MIM:606232)	ARSA (410)	Unsteady gait (HP:0002317)	Gait_disturbance [HS, MP]	MIM:250100
Phelan-Mcdermid syndrome (MIM:606232)	MAPK8IP2 (23542)	Aggressive behavior (HP:0000718)	decreased_anxiety-related_response [MP]	-
Phelan-Mcdermid syndrome (MIM:606232)	MAPK8IP2 (23542)	Aggressive behavior (HP:0000718)	abnormal_social_investigation [MP]	-
Phelan-Mcdermid syndrome (MIM:606232)	MAPK8IP2 (23542)	Aggressive behavior (HP:0000718)	decreased_exploration_in_new_environment [MP]	-
Phelan-Mcdermid syndrome (MIM:606232)	MAPK8IP2 (23542)	Autism (HP:0000717)	decreased_anxiety-related_response [MP]	-
Phelan-Mcdermid syndrome (MIM:606232)	MAPK8IP2 (23542)	Autism (HP:0000717)	decreased_exploration_in_new_environment [MP]	-
Phelan-Mcdermid syndrome (MIM:606232)	MAPK8IP2 (23542)	Poor eye contact (HP:0000817)	abnormal_social_investigation [MP]	-
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Aggressive behavior (HP:0000718)	increased_anxiety-related_response [MP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Aggressive behavior (HP:0000718)	social_withdrawal [MP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Aggressive behavior (HP:0000718)	abnormal_social_investigation [MP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Aggressive behavior (HP:0000718)	increased_aggression_towards_mice [MP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Aggressive behavior (HP:0000718)	abnormal_social/conspecific_interaction [MP]	PMID:20186804
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Autism (HP:0000717)	increased_anxiety-related_response [MP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Broad-based gait (HP:0002136)	abnormal_gait [MP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Dolichocephaly (HP:0000268)	abnormally_decreased_size_head [ZP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Normal to tall stature (HP:0003516)	increased_body_weight [MP]	-
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Poor eye contact (HP:0000817)	social_withdrawal [MP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Poor eye contact (HP:0000817)	abnormal_social_investigation [MP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Poor eye contact (HP:0000817)	abnormal_social/conspecific_interaction [MP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Seizures (HP:0001250)	environmentally_induced_seizures [MP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Stereotypical motor behaviors (HP:0008758)	increased_stereotypic_behavior [MP]	PMID:17173049
Phelan-Mcdermid syndrome (MIM:606232)	SHANK3 (85358)	Unsteady gait (HP:0002317)	abnormal_gait [MP]	PMID:17173049
Potocki-Lupski syndrome (MIM:610883)	ALDH3A2 (224)	Generalized hypotonia (HP:0001290)	Spasticity [HS]	MIM:270200
Potocki-Lupski syndrome (MIM:610883)	ALDH3A2 (224)	Hypermetropia (HP:0000540)	Photophobia [HS]	MIM:270200
Potocki-Lupski syndrome (MIM:610883)	ALDH3A2 (224)	Scoliosis (HP:0002650)	Thoracic_kyphosis [HS]	MIM:270200
Potocki-Lupski syndrome (MIM:610883)	ALDH3A2 (224)	Short stature (HP:0004322)	Short_stature [HS]	MIM:270200
Potocki-Lupski syndrome (MIM:610883)	ATPAF2 (91647)	Generalized hypotonia (HP:0001290)	Muscular_hypotonia [HS]	MIM:604273
Potocki-Lupski syndrome (MIM:610883)	ATPAF2 (91647)	Microcephaly (HP:0000252)	Microcephaly [HS]	MIM:604273
Potocki-Lupski syndrome (MIM:610883)	ATPAF2 (91647)	Micrognathia (HP:0000347)	Retrognathia [HS]	MIM:604273
Potocki-Lupski syndrome (MIM:610883)	ATPAF2 (91647)	Prominent jaw (HP:0002051)	Retrognathia [HS]	MIM:604273
Potocki-Lupski syndrome (MIM:610883)	ATPAF2 (91647)	Prominent nasal tip (HP:0005274)	Prominent_nasal_bridge [HS]	MIM:604273
Potocki-Lupski syndrome (MIM:610883)	ATPAF2 (91647)	Short stature (HP:0004322)	Short_stature [HS]	MIM:604273
Potocki-Lupski syndrome (MIM:610883)	EPN2 (22905)	Low birth weight (HP:0001518)	slow_postnatal_weight_gain [MP]	-
Potocki-Lupski syndrome (MIM:610883)	MAPK7 (5598)	Low birth weight (HP:0001518)	cachexia [MP]	-
Potocki-Lupski syndrome (MIM:610883)	MYO15A (51168)	Echolalia (HP:0010529)	head_bobbing [MP]	-
Potocki-Lupski syndrome (MIM:610883)	MYO15A (51168)	Echolalia (HP:0010529)	circling [MP]	-
Potocki-Lupski syndrome (MIM:610883)	MYO15A (51168)	Stereotypical motor behaviors (HP:0008758)	head_bobbing [MP]	-
Potocki-Lupski syndrome (MIM:610883)	MYO15A (51168)	Stereotypical motor behaviors (HP:0008758)	circling [MP]	-
Potocki-Lupski syndrome (MIM:610883)	PEMT (10400)	Hypocholesterolemia (HP:0003146)	abnormal_lipid_level [MP]	-
Potocki-Lupski syndrome (MIM:610883)	PEMT (10400)	Hypocholesterolemia (HP:0003146)	decreased_circulating_HDL_cholesterol_level [MP]	-
Potocki-Lupski syndrome (MIM:610883)	PEMT (10400)	Hypocholesterolemia (HP:0003146)	decreased_circulating_cholesterol_level [MP]	-
Potocki-Lupski syndrome (MIM:610883)	PEMT (10400)	Hypocholesterolemia (HP:0003146)	abnormal_circulating_cholesterol_level [MP]	-
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Autism (HP:0000717)	increased_anxiety-related_response [MP]	PMID:17357070
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Downslanted palpebral fissures (HP:0000494)	Synophrys [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Echolalia (HP:0010529)	increased_anxiety-related_response [MP]	MIM:182290, PMID:17357070
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Echolalia (HP:0010529)	Stereotyped_repetitiveBehaviour [HS]	MIM:182290, PMID:17357070

Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	EEG abnormality (HP:0002353)	EEG_abnormality [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Gastroesophageal reflux (HP:0002020)	fusion_of_atlas_and_odontoid_process [MP]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Gastroesophageal reflux (HP:0002020)	Abnormality_of_the_thyroid_gland [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Gastroesophageal reflux (HP:0002020)	abnormal_cervical_vertebrae_morphology [MP]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Gastroesophageal reflux (HP:0002020)	abnormal_thyroid_cartilage_morphology [MP]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Generalized hypotonia (HP:0001290)	Generalized_hypotonia [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Hypertelorism (HP:0000316)	Deeply_set_eye [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Hypocholesterolemia (HP:0003146)	increased_circulating_corticosterone_level [MP]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Hypocholesterolemia (HP:0003146)	Hypercholesterolemia [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Hypocholesterolemia (HP:0003146)	Hypertriglyceridemia [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Microcephaly (HP:0000252)	Brachycephaly [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Micrognathia (HP:0000347)	Short_palm [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Poor eye contact (HP:0000817)	Self-mutilation [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Prominent jaw (HP:0002051)	Mandibular_prognathia [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Prominent nasal tip (HP:0005274)	short_snout [MP]	MIM:182290, PMID:17357070
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Prominent nasal tip (HP:0005274)	short_nasal_bone [MP]	MIM:182290, PMID:17357070
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Prominent nasal tip (HP:0005274)	Broad_nasal_bridge [HS]	MIM:182290, PMID:17357070
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Prominent nasal tip (HP:0005274)	broad_nasal_bone [MP]	MIM:182290, PMID:17357070
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Prominent nasal tip (HP:0005274)	abnormal_nasal_bone_morphology [MP]	MIM:182290, PMID:17357070
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Scoliosis (HP:0002650)	kyphosis [MP]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Scoliosis (HP:0002650)	Scoliosis [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Short stature (HP:0004322)	Short_stature [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Sleep apnea (HP:0010535)	Sleep_disturbances [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Speech articulation difficulties (HP:0009088)	Speech_delay [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Stereotypical motor behaviors (HP:0008758)	Stereotyped_repetitiveBehaviour [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Triangular face (HP:0000325)	Malar_hypoplasia [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	RAI1 (10743)	Triangular face (HP:0000325)	Broad_face [HS]	MIM:182290
Potocki-Lupski syndrome (MIM:610883)	SREBF1 (6720)	Hypocholesterolemia (HP:0003146)	abnormal_triglyceride_level [MP]	-
Potocki-Lupski syndrome (MIM:610883)	SREBF1 (6720)	Hypocholesterolemia (HP:0003146)	decreased_circulating_triglyceride_level [MP]	-
Potocki-Lupski syndrome (MIM:610883)	SREBF1 (6720)	Hypocholesterolemia (HP:0003146)	increased_liver_cholesterol_level [MP]	-
Potocki-Lupski syndrome (MIM:610883)	SREBF1 (6720)	Hypocholesterolemia (HP:0003146)	decreased_circulating_cholesterol_level [MP]	-
Potocki-Lupski syndrome (MIM:610883)	SREBF1 (6720)	Hypocholesterolemia (HP:0003146)	increased_circulating_ketone_body_level [MP]	-
Potocki-Lupski syndrome (MIM:610883)	SREBF1 (6720)	Hypocholesterolemia (HP:0003146)	decreased_liver_triglyceride_level [MP]	-
Potocki-Lupski syndrome (MIM:610883)	TOM1L2 (146691)	Autism (HP:0000717)	abnormal_fear/anxiety-related_behavior [MP]	-
Potocki-Lupski syndrome (MIM:610883)	TOM1L2 (146691)	Echolalia (HP:0010529)	abnormal_fear/anxiety-related_behavior [MP]	-
Potocki-Lupski syndrome (MIM:610883)	TOM1L2 (146691)	Hypertelorism (HP:0000316)	exophthalmos [MP]	-
Potocki-Lupski syndrome (MIM:610883)	TOM1L2 (146691)	Hypertelorism (HP:0000316)	abnormal_eye_distance/_position [MP]	-
Potocki-Lupski syndrome (MIM:610883)	TOM1L2 (146691)	Scoliosis (HP:0002650)	kyphosis [MP]	-
Potocki-Lupski syndrome (MIM:610883)	ULK2 (9706)	Autism (HP:0000717)	increased_response_to_stress-induced_hyperthermia [MP]	-
Potocki-Lupski syndrome (MIM:610883)	ULK2 (9706)	Autism (HP:0000717)	increased_anxiety-related_response [MP]	-
Potocki-Lupski syndrome (MIM:610883)	ULK2 (9706)	Echolalia (HP:0010529)	increased_response_to_stress-induced_hyperthermia [MP]	-
Potocki-Lupski syndrome (MIM:610883)	ULK2 (9706)	Echolalia (HP:0010529)	increased_anxiety-related_response [MP]	-
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Brachydactyly (HP:0001156)	polydactyly [MP]	-
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Brachydactyly (HP:0001156)	preaxial_polydactyly [MP]	-
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Broad nasal bridge (HP:0000431)	upturned_snout [MP]	-
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Downslanted palpebral fissures (HP:0000494)	eyelids_open_at_birth [MP]	PMID:19692347
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Epicanthus (HP:0000286)	eyelids_open_at_birth [MP]	-
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Hypoplastic nasal alae (HP:0000430)	small_nasal_bone [MP]	PMID:19692347
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Hypoplastic nasal alae (HP:0000430)	absent_nasal_septum [MP]	PMID:19692347
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Nasal hypoplasia (HP:0003196)	short_snout [MP]	PMID:19692347
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Nasal hypoplasia (HP:0003196)	abnormal_nose_morphology [MP]	PMID:19692347
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Nasal hypoplasia (HP:0003196)	abnormal_nasal_septum_morphology [MP]	PMID:19692347
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Nasal hypoplasia (HP:0003196)	abnormal_nasal_cavity_morphology [MP]	PMID:19692347
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Nasal hypoplasia (HP:0003196)	upturned_snout [MP]	PMID:19692347
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Nasal hypoplasia (HP:0003196)	small_nasal_bone [MP]	PMID:19692347
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Nasal hypoplasia (HP:0003196)	abnormal_nasal_capsule_morphology [MP]	PMID:19692347

Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Nasal hypoplasia (HP:0003196)	absent_nasal_septum [MP]	PMID:19692347
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	short_squamosal_bone [MP]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	small_frontal_bone [MP]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	Parietal_foramina [HS]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	Symmetrical_oval_parietal_bone_defects [HS]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	abnormal_neurocranium_morphology [MP]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	abnormal_fontanelle_morphology [MP]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	abnormal_pterygoid_process_morphology [MP]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	small_aliophenoid_bone [MP]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	abnormal_parietal_bone_morphology [MP]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	abnormal_basisphenoid_bone_morphology [MP]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	small_parietal_bone [MP]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Parietal foramina (HP:0002697)	Parietal_bossing [HS]	PMID:11017806
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Telecanthus (HP:0000506)	eyelids_open_at_birth [MP]	-
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Wormian bones (HP:0002645)	short_squamosal_bone [MP]	PMID:11137991
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Wormian bones (HP:0002645)	small_frontal_bone [MP]	PMID:11137991
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Wormian bones (HP:0002645)	abnormal_neurocranium_morphology [MP]	PMID:11137991
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Wormian bones (HP:0002645)	abnormal_fontanelle_morphology [MP]	PMID:11137991
Potocki-Shaffer syndrome (MIM:601224)	ALX4 (60529)	Wormian bones (HP:0002645)	abnormal_pterygoid_process_morphology [MP]	PMID:11137991
Potocki-Shaffer syndrome (MIM:601224)	CD82 (3732)	Brachydactyly (HP:0001156)	Abnormality_of_the_nail [HS]	-
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Brachydactyly (HP:0001156)	Short_metacarpal [HS]	-
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Hypoplastic nasal alae (HP:0000430)	abnormally_decreased_length_cartilage [ZP]	-
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Hypoplastic nasal alae (HP:0000430)	abnormal_cartilage_morphology [MP]	-
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Hypoplastic nasal alae (HP:0000430)	abnormally_increased_size_Meckel's_cartilage [ZP]	-
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Hypoplastic nasal alae (HP:0000430)	abnormally_decreased_size_ceratobranchial_4_cartilage [ZP]	-
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Hypoplastic nasal alae (HP:0000430)	abnormally_increased_thickness_cartilage [ZP]	-
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Hypoplastic nasal alae (HP:0000430)	abnormally_disrupted_cartilage_development [ZP]	-
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Hypoplastic nasal alae (HP:0000430)	abnormally_deformed_crudal_cartilage [ZP]	-
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Hypoplastic nasal alae (HP:0000430)	abnormally_sloped_downward_Meckel's_cartilage [ZP]	-
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Multiple exostoses (HP:0002762)	Scapular_exostoses [HS]	PMID:9463333
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Multiple exostoses (HP:0002762)	Rib_exostoses [HS]	PMID:9463333
Potocki-Shaffer syndrome (MIM:601224)	EXT2 (2132)	Single transverse palmar crease (HP:0000954)	Short_metacarpal [HS]	MIM:133701
Potocki-Shaffer syndrome (MIM:601224)	SLC35C1 (55343)	Brachycephaly (HP:0000248)	Microcephaly [HS]	MIM:266265
Potocki-Shaffer syndrome (MIM:601224)	SLC35C1 (55343)	Intellectual disability (HP:0001249)	Intellectual_disability_progressive [HS]	PMID:12116250
Potocki-Shaffer syndrome (MIM:601224)	SLC35C1 (55343)	Muscular hypotonia (HP:0001252)	Muscular_hypotonia [HS]	MIM:266265
Potocki-Shaffer syndrome (MIM:601224)	SLC35C1 (55343)	Seizures (HP:0001250)	Seizures [HS]	MIM:266265
Prader-Willi syndrome (MIM:176270)	GABRA5 (2558)	Attention deficit hyperactivity disorder (HP:0007018)	hyperactivity [MP]	PMID:9514592
Prader-Willi syndrome (MIM:176270)	GABRA5 (2558)	Polyphagia (HP:0002591)	decreased_drinking_behavior [MP]	-
Prader-Willi syndrome (MIM:176270)	GABRA5 (2558)	Poor suck (HP:0002033)	decreased_drinking_behavior [MP]	-
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Attention deficit hyperactivity disorder (HP:0007018)	hyperactivity [MP]	-
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Autism (HP:0007017)	abnormal_response_to_novel_object [MP]	PMID:11920158
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Obesity (HP:0001513)	obese [MP]	PMID:12920063
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Oligomenorrhea (HP:0000876)	reduced_female_fertility [MP]	-
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Polyphagia (HP:0002591)	increased_eating_behavior [MP]	PMID:12920063
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Poor suck (HP:0002033)	abnormal_parental_behavior [MP]	-
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Poor suck (HP:0002033)	abnormal_maternal_nurturing [MP]	-
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Poor suck (HP:0002033)	reduced_male_mating_frequency [MP]	-
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Poor suck (HP:0002033)	abnormal_social/conspicuous_interaction [MP]	-
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Poor suck (HP:0002033)	abnormal_nest_building_behavior [MP]	-
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Primary amenorrhea (HP:0000786)	reduced_female_fertility [MP]	-
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Seizures (HP:0001250)	seizures [MP]	PMID:19533781
Prader-Willi syndrome (MIM:176270)	GABRB3 (2562)	Seizures (HP:0001250)	absence_seizures [MP]	PMID:19533781
Prader-Willi syndrome (MIM:176270)	HERC2 (8924)	Clitoral hypoplasia (HP:0000060)	decreased_corpora_lutea_number [MP]	-
Prader-Willi syndrome (MIM:176270)	HERC2 (8924)	Hypoplastic labia minora (HP:0000064)	decreased_corpora_lutea_number [MP]	-
Prader-Willi syndrome (MIM:176270)	HERC2 (8924)	Infertility (HP:0000789)	male_infertility [MP]	PMID:9949213
Prader-Willi syndrome (MIM:176270)	HERC2 (8924)	Infertility (HP:0000789)	female_infertility [MP]	PMID:9949213
Prader-Willi syndrome (MIM:176270)	HERC2 (8924)	Oligomenorrhea (HP:0000876)	reduced_female_fertility [MP]	PMID:9949213
Prader-Willi syndrome (MIM:176270)	HERC2 (8924)	Primary amenorrhea (HP:0000786)	reduced_female_fertility [MP]	PMID:9949213
Prader-Willi syndrome (MIM:176270)	NDN (4692)	Generalized hypotonia (HP:0001290)	hypotonia [MP]	-
Prader-Willi syndrome (MIM:176270)	NDN (4692)	Hypoventilation (HP:0002791)	hypoventilation [MP]	PMID:10508517
Prader-Willi syndrome (MIM:176270)	NDN (4692)	Hypoventilation (HP:0002791)	respiratory_distress [MP]	PMID:10508517
Prader-Willi syndrome (MIM:176270)	NDN (4692)	Hypoventilation (HP:0002791)	respiratory_distress [MP]	PMID:10508517
Prader-Willi syndrome (MIM:176270)	NIPA1 (123606)	Sleep apnea (HP:0010533)	Spastic_gait [HS]	PMID:15711826
Prader-Willi syndrome (MIM:176270)	NIPA1 (123606)	Generalized hypotonia (HP:0001290)	Spastic_paraplegia [HS]	PMID:15711826
Prader-Willi syndrome (MIM:176270)	NIPA1 (123606)	Generalized hypotonia (HP:0001290)	Lower_limb_spasticity [HS]	PMID:15711826
Prader-Willi syndrome (MIM:176270)	NIPA1 (123606)	Small feet (HP:0001290)	Pes_cavus [HS]	PMID:15711826
Prader-Willi syndrome (MIM:176270)	NIPA1 (123606)	Small feet (HP:0001764)	Lower_limb_muscle_weakness [HS]	PMID:15711826
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Attention deficit hyperactivity disorder (HP:0007018)	nervous [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Cryptorchidism (HP:0000028)	abnormal_sperm_head_morphology [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Cryptorchidism (HP:0000028)	enlarged_sperm_head [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Cryptorchidism (HP:0000028)	abnormal_acrosome_morphology [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Cryptorchidism (HP:0000028)	small_seminiferous_tubules [MP]	-

Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hyperinsulinemia (HP:0000842)	hyperglycemia [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hyperinsulinemia (HP:0000842)	abnormal glucose homeostasis [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hyperinsulinemia (HP:0000842)	increased_circulating_insulin_level [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypermetropia (HP:0000540)	Impaired_vision [HS]	PMID:18680187
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	variegated_coat_color [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	yellow_coat_color [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	abnormal_coat_hair_pigmentation [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	mottled_coat [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	delayed_hair_regrowth [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	abnormal_melanosome_morphology [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	abnormal_hair_follicle_melanin_granule_morphology [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	abnormal_hair_follicle_melanin_granule_distribution [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	abnormal_melanogenesis [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	abnormal_hair_follicle_melanin_granule_shape [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	darkened_coat_color [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	premature_hair_loss [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	Red_hair [HS]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of hair (HP:0005599)	diluted_coat_color [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	abnormal_ear_pigmentation [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	decreased_tail_pigmentation [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	Freckling [HS]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	abnormal_melanosome_morphology [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	abnormal_melanogenesis [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	abnormal_melanosome_morphology [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	abnormal_skin_pigmentation [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	Freckles_in_sun-exposed_areas [HS]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypopigmentation of the skin (HP:0001010)	reduced_eye_pigmentation [MP]	PMID:1509264
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Hypoplasia of penis (HP:0008736)	testis_hypoplasia [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Infertility (HP:0000789)	infertility [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Infertility (HP:0000789)	female_infertility [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Infertility (HP:0000789)	male_infertility [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Obesity (HP:0001513)	increased_body_weight [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Poor suck (HP:0002033)	abnormal_pup_retrieval [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Poor suck (HP:0002033)	abnormal_maternal_nursuring [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Recurrent respiratory infections (HP:0002205)	increased_susceptibility_to_bacterial_infection [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Reduced iris pigmentation (HP:0007730)	ocular_albinism [MP]	PMID:8302318
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Reduced iris pigmentation (HP:0007730)	Albinism [HS]	PMID:8302318
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Reduced iris pigmentation (HP:0007730)	abnormal_melanosome_morphology [MP]	PMID:8302318
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Reduced iris pigmentation (HP:0007730)	abnormal_melanogenesis [MP]	PMID:8302318
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Reduced iris pigmentation (HP:0007730)	abnormal_choroid_melanin_granule_morphology [MP]	PMID:8302318
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Reduced iris pigmentation (HP:0007730)	absent_eye_pigmentation [MP]	PMID:8302318
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Reduced iris pigmentation (HP:0007730)	abnormal_eye_pigmentation [MP]	PMID:8302318
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Scrotal hypoplasia (HP:0000046)	testis_hypoplasia [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Type II diabetes mellitus (HP:0005978)	hyperglycemia [MP]	-
Prader-Willi syndrome (MIM:176270)	OCA2 (4948)	Type II diabetes mellitus (HP:0005978)	abnormal_glucose_homeostasis [MP]	-
Prader-Willi syndrome (MIM:176270)	SNRPN (6638)	Hyperinsulinemia (HP:0000842)	decreased_circulating_glucose_level [MP]	-
Prader-Willi syndrome (MIM:176270)	SNRPN (6638)	Obesity (HP:0001513)	increased_body_weight [MP]	-
Prader-Willi syndrome (MIM:176270)	SNRPN (6638)	Obesity (HP:0001513)	increased_body_size [MP]	-
Prader-Willi syndrome (MIM:176270)	SNRPN (6638)	Polyphagia (HP:0002591)	absent_gastric_milk_in_neonates [MP]	-
Prader-Willi syndrome (MIM:176270)	SNRPN (6638)	Poor suck (HP:0002033)	abnormal_suckling_behavior [MP]	-
Prader-Willi syndrome (MIM:176270)	SNRPN (6638)	Type II diabetes mellitus (HP:0005978)	decreased_circulating_glucose_level [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Attention deficit hyperactivity disorder (HP:0007018)	Hyperactivity [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Citral hypoplasia (HP:0000060)	decreased_uterus_weight [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Citral hypoplasia (HP:0000060)	abnormal_mammary_gland_growth_during_pregnancy [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Citral hypoplasia (HP:0000060)	decreased_ovary_weight [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Cryptorchidism (HP:0000028)	decreased_prostate_gland_weight [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Dolichocephaly (HP:0000268)	Microcephaly,_postnatal [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Dolichocephaly (HP:0000268)	Flat_occiput [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Dolichocephaly (HP:0000268)	Microbrachycephaly [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Feeding problems in infancy (HP:0008872)	Feeding_difficulties [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Generalized hypotonia (HP:0001290)	Muscular_hypotonia [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Hypopigmentation of hair (HP:0005599)	Blond_hair [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Hypopigmentation of the skin (HP:0001010)	Hypopigmentation_of_the_skin [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Hypoplasia of penis (HP:0008736)	decreased_testis_weight [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Hypoplastic labia minora (HP:0000064)	decreased_uterus_weight [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Hypoplastic labia minora (HP:0000064)	abnormal_mammary_gland_growth_during_pregnancy [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Hypoplastic labia minora (HP:0000064)	decreased_ovary_weight [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Impaired language development (HP:0000750)	Absent_speech_development [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Motor delay (HP:0001270)	Motor_delay [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Obesity (HP:0001513)	Obesity [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Oligomenorrhea (HP:0000876)	abnormal_ovarian_folliculogenesis [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Oligomenorrhea (HP:0000876)	impaired_luteinization [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Oligomenorrhea (HP:0000876)	reduced_female_fertility [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Poor suck (HP:0002033)	Feeding_difficulties [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Poor suck (HP:0002033)	Paroxysmal_bursts_of_laughter [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Primary amenorrhea (HP:000786)	abnormal_ovarian_folliculogenesis [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Primary amenorrhea (HP:000786)	impaired_luteinization [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Primary amenorrhea (HP:000786)	reduced_female_fertility [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Reduced iris pigmentation (HP:0007730)	Blue_irides [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Scoliosis (HP:0002650)	Scoliosis [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Scrotal hypoplasia (HP:0000046)	decreased_testis_weight [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Seizures (HP:0001250)	tonic-clonic_seizures [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Seizures (HP:0001250)	audiovisual_seizures [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Seizures (HP:0001250)	Seizures [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Seizures (HP:0001250)	absence_seizures [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Seizures (HP:0001250)	abnormal_spike_wave_discharge [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Short palm (HP:0004279)	Hypoplasia_of_the_maxilla [HS]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Sleep apnea (HP:0010535)	abnormal_sleep_pattern [MP]	-
Prader-Willi syndrome (MIM:176270)	UBE3A (7337)	Sleep apnea (HP:0010535)	Sleep-wake_cycle_disturbance [HS]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Abnormal liver function tests (HP:0001411)	abnormal_liver_physiology [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Abnormality of alkaline phosphatase activity (HP:0004379)	abnormal_enzyme/coenzyme_activity [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Abnormality of alkaline phosphatase activity (HP:0004379)	increased_circulating_aspartate_transaminase_level [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Glucose intolerance (HP:0000833)	abnormal_glucose_homeostasis [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Glucose intolerance (HP:0000833)	increased_circulating_insulin_level [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Glucose intolerance (HP:0000833)	hyperglycemia [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Glycosuria (HP:0003076)	abnormal_glucose_homeostasis [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Glycosuria (HP:0003076)	increased_circulating_insulin_level [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Glycosuria (HP:0003076)	hyperglycemia [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Insulin-dependent maturity-onset diabetes of the young (HP:0004904)	abnormal_glucose_homeostasis [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Insulin-dependent maturity-onset diabetes of the young (HP:0004904)	increased_circulating_insulin_level [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	ACACA (31)	Insulin-dependent maturity-onset diabetes of the young (HP:0004904)	hyperglycemia [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Abnormal liver function tests (HP:0001411)	liver_inflammation [MP]	PMID:15068978

RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Abnormal liver function tests (HP:0001411)	abnormal_hepatobiliary_system_physiology [MP]	PMID:15068978
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Abnormal liver function tests (HP:0001411)	decreased_liver_function [MP]	PMID:15068978
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Biliary tract abnormality (HP:0001080)	abnormal_intrahepatic_bile_duct_morphology [MP]	PMID:15001636
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Biliary tract abnormality (HP:0001080)	abnormal_extrahepatic_bile_duct_morphology [MP]	PMID:15001636
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Biliary tract abnormality (HP:0001080)	jaundice [MP]	PMID:15001636
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Biliary tract abnormality (HP:0001080)	abnormal_interlobular_bile_duct_morphology [MP]	PMID:15001636
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Biliary tract abnormality (HP:0001080)	abnormal_cystic_duct_morphology [MP]	PMID:15001636
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Biliary tract abnormality (HP:0001080)	intraluminal_cholestasis [MP]	PMID:15001636
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Biliary tract abnormality (HP:0001080)	abnormal_bile_duct_physiology [MP]	PMID:15001636
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Biliary tract abnormality (HP:0001080)	abnormal_gallbladder_morphology [MP]	PMID:15001636
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Exocrine pancreatic insufficiency (HP:0001738)	abnormally_quality_endocrine_pancreas [ZP]	PMID:15068978
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Glucose intolerance (HP:0000833)	Type_II_diabetes_mellitus [HS]	MIM:125853
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Glycosuria (HP:0003076)	Type_II_diabetes_mellitus [HS]	MIM:125853
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Gout (HP:0001997)	increased_circulating_bilirubin_level [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Increased creatinine (HP:0003259)	increased_circulating_bilirubin_level [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Insulin-dependent maturity-onset diabetes of the young	Type_II_diabetes_mellitus [HS]	MIM:125853
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Insulin-dependent maturity-onset diabetes of the young	insulin_resistance [HS]	MIM:125853
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Pancreatic hypoplasia (HP:0002594)	abnormally_quality_endocrine_pancreas [ZP]	PMID:15068978
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Pancreatic hypoplasia (HP:0002594)	abnormally_aplastic_pancreas [ZP]	PMID:15068978
RCAD (renal cysts and diabetes) (MIM:137920)	HNF1B (6928)	Pancreatic hypoplasia (HP:0002594)	abnormally_disrupted_pancreas_development [ZP]	PMID:15068978
RCAD (renal cysts and diabetes) (MIM:137920)	LHX1 (3975)	Hypoplastic glomerulocystic kidney disease (HP:0100611)	decreased_nephron_number [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	LHX1 (3975)	Hypoplastic glomerulocystic kidney disease (HP:0100611)	absent_nephron [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	LHX1 (3975)	Hypoplastic glomerulocystic kidney disease (HP:0100611)	absent_renal_glomerulus [MP]	-
RCAD (renal cysts and diabetes) (MIM:137920)	LHX1 (3975)	Hypoplastic glomerulocystic kidney disease (HP:0100611)	decreased_renal_glomerulus_number [MP]	-
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Atrial septal defect (HP:0001631)	atrial_septal_defect [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Beaked nose (HP:0000444)	nasal_bone_hypoplasia [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Beaked nose (HP:0000444)	short_snout [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Beaked nose (HP:0000444)	broad_nasal_bridge [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Broad hallux (HP:0010055)	oligodactyly [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Broad thumb (HP:0011304)	oligodactyly [MP]	-
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Broad thumb (HP:0011304)	abnormal_rib_morphology [MP]	-
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Broad thumb (HP:0011304)	xiphoid_process_foramen [MP]	-
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Broad thumb (HP:0011304)	abnormal_sternum_morphology [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Capillary hemangiomas (HP:0005306)	leukemia [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Capillary hemangiomas (HP:0005306)	T_cell_derived_lymphoma [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Capillary hemangiomas (HP:0005306)	increased_histiocytic_sarcoma_incidence [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Capillary hemangiomas (HP:0005306)	increased_hemolymphoid_system_tumor_incidence [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Clinodactyly of the 5th finger (HP:0004209)	oligodactyly [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Columnella, low hanging (HP:0009765)	broad_nasal_bridge [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Deviated nasal septum (HP:0004411)	broad_nasal_bridge [MP]	PMID:18792986
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	High-arched palate (HP:0000156)	cleft_secondary_palate [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Hypoplasia of the maxilla (HP:0000327)	small_maxilla [MP]	-
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Hypoplasia of the maxilla (HP:0000327)	short_premaxilla [MP]	-
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Hypoplasia of the maxilla (HP:0000327)	short_maxilla [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Patent ductus arteriosus (HP:0001643)	abnormal_vascular_endothelial_cell_development [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Patent ductus arteriosus (HP:0001643)	abnormal_vascular_development [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Patent ductus arteriosus (HP:0001643)	absent_organized_vascular_network [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Patent ductus arteriosus (HP:0001643)	failure_of_vascular_branching [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Patent ductus arteriosus (HP:0001643)	abnormal_vasculogenesis [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Patent ductus arteriosus (HP:0001643)	decreased_angiogenesis [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Patent ductus arteriosus (HP:0001643)	absent_vitelline_blood_vessels [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Prominent fingertip pads (HP:0001212)	oligodactyly [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Radial deviation of thumb terminal phalanx (HP:0005895)	oligodactyly [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Recurrent upper respiratory tract infections (HP:0002788)	broad_nasal_bridge [MP]	PMID:18792986
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Scoliosis (HP:0002650)	scoliosis [MP]	PMID:9294190
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Seizures (HP:0001250)	seizures [MP]	PMID:16868563

Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Small, flared iliac wings (HP:0003181)	xiphoid_process_foramen [MP]	-
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Small, flared iliac wings (HP:0003181)	abnormal_sternum_morphology [MP]	PMID:18792986
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Spina bifida occulta (HP:0003298)	abnormal_neural_tube_morphology/development [MP]	-
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Spina bifida occulta (HP:0003298)	abnormal_neural_tube_closure [MP]	-
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Spina bifida occulta (HP:0003298)	open_neural_tube [MP]	-
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Ventricular septal defect (HP:0001629)	perimembranous_ventricular_septal_defect [MP]	PMID:16868563
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Wide anterior fontanel (HP:0000260)	large_anterior_fontanelle [MP]	PMID:9294190
Rubinstein-Taybi syndrome (MIM:180849)	CREBBP (1387)	Wide anterior fontanel (HP:0000260)	wide_frontal_bone [MP]	PMID:9294190
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_crista_ampullaris_neuroepithelium_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_outer_ear_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_inner_ear_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_malleus_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal Cochlea_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_vestibule_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	absent_utricle [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_otic_capsule_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	absent_semicircular_canals [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	otic_capsule_hypoplasia [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	small_otic_capsule [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	absent_endolymphatic_duct [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_middle_ear_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_stapedial_artery_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	short_endolymphatic_duct [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	absent_stapes [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	decreased Cochlear_coiling [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	absent_superior_semicircular_canal [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	absent_posterior_semicircular_canal [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	decreased_lateral_semicircular_canal_size [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_vestibular_saccule_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_crista_ampullaris_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_scala_vestibuli_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	enlarged_utricle [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_utricular_macula_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_vestibular_saccular_macula_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_external_auditory_canal_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	decreased_tympanic_ring_size [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_tympanic_membrane_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_otic_vesicle_development [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_middle_ear_ossicle_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	small_malleus_processus_brevis [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	absent_malleus_head [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_gonial_bone_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	absent_vestibular_saccule [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	absent_vestibule [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	abnormal_semicircular_canal_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	otic_vesicle_hypoplasia [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Abnormality of the pinna (HP:0000377)	small_otic Vesicle [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Cleft palate (HP:0000175)	abnormal_palate_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Cleft palate (HP:0000175)	cleft_primary_palate [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Cleft palate (HP:0000175)	palatal_shelves_fail_to_meet_at_midline [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Cleft palate (HP:0000175)	cleft_secondary_palate [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Cleft palate (HP:0000175)	decreased_maxillary_shelf_size [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Cleft palate (HP:0000175)	absent_palatine_shelf [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Ectrodactyly (feet) (HP:0001839)	abnormal_hindlimb_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Ectrodactyly (feet) (HP:0001839)	monodactyly [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX5 (1749)	Ectrodactyly (feet) (HP:0001839)	brachydactyly [MP]	PMID:12000792

Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Ectrodactyly (feet) (HP:0001839)	ectrodactyly [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Ectrodactyly (feet) (HP:0001839)	abnormal_digit_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Ectrodactyly (feet) (HP:0001839)	abnormal_metatarsal_bone_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Ectrodactyly (feet) (HP:0001839)	abnormal_phalanx_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Ectrodactyly (hands) (HP:0001171)	monodactyly [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Ectrodactyly (hands) (HP:0001171)	brachydactyly [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Ectrodactyly (hands) (HP:0001171)	ectrodactyly [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Ectrodactyly (hands) (HP:0001171)	abnormal_digit_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Ectrodactyly (hands) (HP:0001171)	abnormal_phalanx_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (feet) (HP:0001849)	abnormal_hindlimb_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (feet) (HP:0001849)	monodactyly [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (feet) (HP:0001849)	brachydactyly [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (feet) (HP:0001849)	mandible_hypoplasia [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (feet) (HP:0001849)	abnormal_digit_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (feet) (HP:0001849)	abnormal_metatarsal_bone_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (feet) (HP:0001849)	abnormal_phalanx_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (hands) (HP:0001180)	abnormal_hindlimb_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (hands) (HP:0001180)	monodactyly [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (hands) (HP:0001180)	brachydactyly [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (hands) (HP:0001180)	mandible_hypoplasia [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (hands) (HP:0001180)	abnormal_digit_morphology [MP]	PMID:12000792
Split hand/foot malformation 1 (MIM:183600)	DLX6 (1750)	Oligodactyly (hands) (HP:0001180)	abnormal_phalanx_morphology [MP]	PMID:12000792
Smith-Magenis syndrome (MIM:182290)	ALDH3A2 (224)	Abnormality of dental morphology (HP:0006482)	Hypoplasia_of_dental_enamel [HS]	MIM:270200
Smith-Magenis syndrome (MIM:182290)	ALDH3A2 (224)	Brachydactyly (HP:0001156)	Abnormality_of_the_hand [HS]	MIM:270200
Smith-Magenis syndrome (MIM:182290)	ALDH3A2 (224)	Brachydactyly (HP:0001156)	Abnormality_of_the_nail [HS]	MIM:270200
Smith-Magenis syndrome (MIM:182290)	ALDH3A2 (224)	Broad palm (HP:0001169)	Abnormality_of_the_hand [HS]	MIM:270200
Smith-Magenis syndrome (MIM:182290)	ALDH3A2 (224)	Generalized hypotonia (HP:0001290)	Spasticity [HS]	MIM:270200
Smith-Magenis syndrome (MIM:182290)	ALDH3A2 (224)	Pes planus (HP:0001763)	Abnormality_of_the_feet [HS]	MIM:270200
Smith-Magenis syndrome (MIM:182290)	ALDH3A2 (224)	Scoliosis (HP:0002650)	Thoracic_kyphosis [HS]	MIM:270200
Smith-Magenis syndrome (MIM:182290)	ALDH3A2 (224)	Seizures (HP:0001250)	Seizures [HS]	MIM:270200
Smith-Magenis syndrome (MIM:182290)	ALDH3A2 (224)	Short palm (HP:0004279)	Abnormality_of_the_hand [HS]	MIM:270200
Smith-Magenis syndrome (MIM:182290)	ALDH3A2 (224)	Short stature (HP:0004322)	Short_stature [HS]	MIM:270200
Smith-Magenis syndrome (MIM:182290)	ALDH3A2 (224)	Synophrys (HP:0000664)	Abnormality_of_the_hair [HS]	MIM:270200
Smith-Magenis syndrome (MIM:182290)	ATPAF2 (91647)	Abnormality of the outer ear (HP:0000356)	Low-set_ears [HS]	MIM:604273
Smith-Magenis syndrome (MIM:182290)	ATPAF2 (91647)	Brachycephaly (HP:0000248)	Microcephaly [HS]	MIM:604273
Smith-Magenis syndrome (MIM:182290)	ATPAF2 (91647)	Broad nasal bridge (HP:0000431)	Prominent_nasal_bridge [HS]	MIM:604273
Smith-Magenis syndrome (MIM:182290)	ATPAF2 (91647)	Generalized hypotonia (HP:0001290)	Muscular_hypotonia [HS]	MIM:604273
Smith-Magenis syndrome (MIM:182290)	ATPAF2 (91647)	Mandibular prognathia (HP:0000303)	Retrognathia [HS]	MIM:604273
Smith-Magenis syndrome (MIM:182290)	ATPAF2 (91647)	Short stature (HP:0004322)	Short_stature [HS]	MIM:604273
Smith-Magenis syndrome (MIM:182290)	B9D1 (27077)	Brachydactyly (HP:0001156)	polydactyly [MP]	-
Smith-Magenis syndrome (MIM:182290)	B9D1 (27077)	Brachydactyly (HP:0001156)	preaxial_polydactyly [MP]	-
Smith-Magenis syndrome (MIM:182290)	FLCN (201163)	Synophrys (HP:0000664)	Abnormality_of_the_hair [HS]	MIM:240500
Smith-Magenis syndrome (MIM:182290)	LLGL1 (3996)	Brachycephaly (HP:0000248)	domed_cranium [MP]	-
Smith-Magenis syndrome (MIM:182290)	LLGL1 (3996)	Ventriculomegaly (HP:0002119)	hydroencephaly [MP]	PMID:8565641
Smith-Magenis syndrome (MIM:182290)	LLGL1 (3996)	Ventriculomegaly (HP:0002119)	abnormal_brain_ventricle_morphology [MP]	PMID:8565641
Smith-Magenis syndrome (MIM:182290)	LLGL1 (3996)	Ventriculomegaly (HP:0002119)	intraventricular_hemorrhage [MP]	PMID:8565641
Smith-Magenis syndrome (MIM:182290)	LLGL1 (3996)	Ventriculomegaly (HP:0002119)	enlarged_lateral_ventricles [MP]	PMID:8565641
Smith-Magenis syndrome (MIM:182290)	MAPK7 (5598)	Abnormality of dental morphology (HP:0006482)	small_first_branchial_arch [MP]	-
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	increased_susceptibility_to_age-related_hearing_loss [MP]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	abnormal_hearing_electrophysiology [MP]	MIM:600316, PMID:11735029

Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	Bilateral_sensorineural_deafness [HS]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	abnormal_hair_cell_physiology [MP]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	absent_cochlear_microphonics [MP]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	absent_distortion_product_otoacoustic_emissions [MP]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	deafness [MP]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	decreased_brainstem_auditory_evoked_potential [MP]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	abnormal_vestibular_system_physiology [MP]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	absent_brainstem_auditory_evoked_potential [MP]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	Congenital_sensorineural_deafness [HS]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	abnormal_hair_cell_mechanolectric_transduction [MP]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Hearing impairment (HP:0000365)	impaired_hearing [MP]	MIM:600316, PMID:11735029
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	short_inner_hair_cell_stereocilia [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	absent Cochlear_hair_cells [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	decreased_outer_hair_cell_stereocilia_number [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	short_outer_hair_cell_stereocilia [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	abnormal_inner_hair_cell_stereociliary_bundle_morphology [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	decreased_inner_hair_cell_stereocilia_number [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	abnormal_cochlear_hair_cell_stereociliary_bundle_morphology [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	short_cochlear_hair_cell_stereocilia [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	Bilateral_conductive_deafness [HS]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	abnormal_outer_hair_cell_stereociliary_bundle_morphology [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	abnormal_cochlear_hair_bundle_tip_links_morphology [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	abnormal_cochlear_outer_hair_cell_morphology [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Middle ear malformations (HP:0008609)	abnormal_organ_of_Corti_morphology [MP]	MIM:600316
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Stereotyped, repetitive behaviour (HP:0000733)	head_bobbing [MP]	-
Smith-Magenis syndrome (MIM:182290)	MYO15A (51168)	Stereotyped, repetitive behaviour (HP:0000733)	circling [MP]	-
Smith-Magenis syndrome (MIM:182290)	PEMT (10400)	Hypercholesterolemia (HP:0003124)	abnormal_lipid_level [MP]	-
Smith-Magenis syndrome (MIM:182290)	PEMT (10400)	Hypercholesterolemia (HP:0003124)	decreased_circulating_HDL_cholesterol_level [MP]	-
Smith-Magenis syndrome (MIM:182290)	PEMT (10400)	Hypercholesterolemia (HP:0003124)	decreased_circulating_cholesterol_level [MP]	-
Smith-Magenis syndrome (MIM:182290)	PEMT (10400)	Hypercholesterolemia (HP:0003124)	abnormal_circulating_cholesterol_level [MP]	-
Smith-Magenis syndrome (MIM:182290)	PEMT (10400)	Hypertriglyceridemia (HP:0002155)	abnormal_lipid_level [MP]	-
Smith-Magenis syndrome (MIM:182290)	PEMT (10400)	Hyporeflexia (HP:0001265)	impaired_righting_response [MP]	-
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Abnormality of dental morphology (HP:0006482)	malocclusion [MP]	-
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Abnormality of the thyroid gland (HP:0000820)	fusion_of_atlas_and_odontoid_process [MP]	-
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Abnormality of the thyroid gland (HP:0000820)	abnormal_cervical_vertebrae_morphology [MP]	-
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Brachydactyly (HP:0001156)	polyphalangy [MP]	PMID:15788730
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Broad nasal bridge (HP:0000431)	broad_nasal_bone [MP]	PMID:16845274
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Broad nasal bridge (HP:0000431)	abnormal_snout_morphology [MP]	PMID:16845274
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Broad nasal bridge (HP:0000431)	short_snout [MP]	PMID:16845274
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Broad nasal bridge (HP:0000431)	short_nasal_bone [MP]	PMID:16845274
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Broad nasal bridge (HP:0000431)	abnormal_nasal_bone_morphology [MP]	PMID:16845274
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Hoarse voice (HP:0001609)	abnormal_thyroid_cartilage_morphology [MP]	PMID:15788730
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Hyperactivity (HP:0000752)	hyperactivity [MP]	PMID:15788730
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Hypercholesterolemia (HP:0003124)	increased_circulating_corticosterone_level [MP]	-
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Hypertriglyceridemia (HP:0002155)	increased_circulating_corticosterone_level [MP]	-
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Scoliosis (HP:0002650)	kyphosis [MP]	PMID:15788730
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Seizures (HP:0001250)	tonic-clonic_seizures [MP]	PMID:15788730
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Seizures (HP:0001250)	seizures [MP]	PMID:15788730
Smith-Magenis syndrome (MIM:182290)	RAI1 (10743)	Self-mutilation (HP:0000742)	increased_anxiety-related_response [MP]	PMID:15788730
Smith-Magenis syndrome (MIM:182290)	SREBF1 (6720)	Hypercholesterolemia (HP:0003124)	increased_liver_cholesterol_level [MP]	PMID:12180145
Smith-Magenis syndrome (MIM:182290)	SREBF1 (6720)	Hypercholesterolemia (HP:0003124)	decreased_circulating_cholesterol_level [MP]	PMID:12180145
Smith-Magenis syndrome (MIM:182290)	SREBF1 (6720)	Hypercholesterolemia (HP:0003124)	increased_circulating_ketone_body_level [MP]	PMID:12180145
Smith-Magenis syndrome (MIM:182290)	SREBF1 (6720)	Hypertriglyceridemia (HP:0002155)	abnormal_triglyceride_level [MP]	PMID:12180145
Smith-Magenis syndrome (MIM:182290)	SREBF1 (6720)	Hypertriglyceridemia (HP:0002155)	decreased_circulating_triglyceride_level [MP]	PMID:12180145
Smith-Magenis syndrome (MIM:182290)	SREBF1 (6720)	Hypertriglyceridemia (HP:0002155)	increased_circulating_ketone_body_level [MP]	PMID:12180145
Smith-Magenis syndrome (MIM:182290)	SREBF1 (6720)	Hypertriglyceridemia (HP:0002155)	decreased_liver_triglyceride_level [MP]	PMID:12180145

Smith-Magenis syndrome (MIM:182290)	TNFRSF13B (23495)	Abnormality of the tracheobronchial system (HP:0005607)	Bronchiectasis [HS]	MIM:240500
Smith-Magenis syndrome (MIM:182290)	TNFRSF13B (23495)	Constipation (HP:0002019)	Diarrhea [HS]	MIM:240500
Smith-Magenis syndrome (MIM:182290)	TNFRSF13B (23495)	Middle ear malformations (HP:0008609)	Acute_otoitis_media [HS]	MIM:240500
Smith-Magenis syndrome (MIM:182290)	TOM1L2 (146691)	Abnormality of dental morphology (HP:0006482)	malocclusion [MP]	-
Smith-Magenis syndrome (MIM:182290)	TOM1L2 (146691)	Deeply set eye (HP:0000490)	exophthalmos [MP]	-
Smith-Magenis syndrome (MIM:182290)	TOM1L2 (146691)	Deeply set eye (HP:0000490)	abnormal_eye_distance/_position [MP]	-
Smith-Magenis syndrome (MIM:182290)	TOM1L2 (146691)	Hyperactivity (HP:0000752)	hyperactivity [MP]	-
Smith-Magenis syndrome (MIM:182290)	TOM1L2 (146691)	Scoliosis (HP:0002650)	kyphosis [MP]	-
Smith-Magenis syndrome (MIM:182290)	TOM1L2 (146691)	Self-mutilation (HP:0000742)	abnormal_fear/anxiety-related_behavior [MP]	-
Smith-Magenis syndrome (MIM:182290)	TOM1L2 (146691)	Synophris (HP:0000664)	focal_hair_loss [MP]	-
Smith-Magenis syndrome (MIM:182290)	TOM1L2 (146691)	Ventriculomegaly (HP:0002119)	hydroencephaly [MP]	-
Smith-Magenis syndrome (MIM:182290)	ULK2 (9706)	Self-mutilation (HP:0000742)	increased_response_to_stress-induced_hyperthermia [MP]	-
Smith-Magenis syndrome (MIM:182290)	ULK2 (9706)	Self-mutilation (HP:0000742)	increased_anxiety-related_response [MP]	-
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Dolichocephaly (HP:0000268)	Macrocephaly [HS]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Dolichocephaly (HP:0000268)	abnormally_decreased_size_head [ZP]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	High anterior hairline (HP:0009890)	Sparse_scalp_hair [HS]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Joint laxity (HP:0001388)	Joint_laxity [HS]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Large feet (HP:0001833)	Pes_planus [HS]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Large feet (HP:0001833)	Coxa_valga [HS]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Large feet (HP:0001833)	Cutis_gyrata_of_palms_and_soles [HS]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Large feet (HP:0001833)	Single_transverse_palmar_cleft [HS]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Large feet (HP:0001833)	Arachnodactyly [HS]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Large hands (HP:0001176)	Single_transverse_palmar_cleft [HS]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Macrocephaly (HP:0000256)	Macrocephaly [HS]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Macrocephaly (HP:0000256)	abnormally_decreased_size_head [ZP]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Neonatal hypotonia (HP:0001319)	Muscular_hypotonia [HS]	MIM:130070
Sotos syndrome (MIM:117550)	B4GALT7 (11285)	Tall stature (HP:0000098)	Short_stature [HS]	MIM:130070
Sotos syndrome (MIM:117550)	CPLX2 (10814)	Partial agenesis of the corpus callosum (HP:0001338)	small_hippocampus [MP]	-
Sotos syndrome (MIM:117550)	NSD1 (64324)	Accelerated skeletal maturation (HP:0005616)	Accelerated_skeletal_maturation [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Accelerated skeletal maturation (HP:0005616)	Dysharmonic_bone_age [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Cavum septum pellucidum (HP:0002389)	Absent_septum_pellucidum [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Dolichocephaly (HP:0000268)	Macrocephaly [HS]	MIM:130650, MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Dolichocephaly (HP:0000268)	Prominent_occiput [HS]	MIM:130650, MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Downslanted palpebral fissures (HP:0000494)	Epicanthus [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Downslanted palpebral fissures (HP:0000494)	Downslanted_palpebral_fissures [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Expressive language delay (HP:0002474)	Speech_delay [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Frontal bossing (HP:0002007)	Prominent_occiput [HS]	MIM:130650, MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	High anterior hairline (HP:0009890)	Thin_hair [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	High anterior hairline (HP:0009890)	Thin_deep-set_nails [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Joint laxity (HP:0001388)	Limited_elbow_extension [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Talipes_equinovarus [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Pes_cavus [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Hypoplastic_iliac_wings [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Joint_contractures_involving_the_joints_of_the_hand [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Broad_thumb [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Disharmonious_carpal_bone [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Coxa_valga [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Calcanevalgus_deformity [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Overlapping_toes [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Metatarsus_adductus [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Short_fourth_metatarsal [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Limited_knee_extension [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Radial_deviation_of_finger [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large feet (HP:0001833)	Prominent_fingertip_pads [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large hands (HP:0001176)	Joint_contractures_involving_the_joints_of_the_hand [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large hands (HP:0001176)	Broad_thumb [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large hands (HP:0001176)	Disharmonious_carpal_bone [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large hands (HP:0001176)	Large_hands [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large hands (HP:0001176)	Radial_deviation_of_finger [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Large hands (HP:0001176)	Prominent_fingertip_pads [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Macrocephaly (HP:0000256)	Macrocephaly [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Neonatal hypotonia (HP:0001319)	Muscular_hypotonia [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Prominent jaw (HP:0002051)	Retrognathia [HS]	MIM:130650, MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Prominent jaw (HP:0002051)	Prominent_chin_with_central_dimple [HS]	MIM:130650, MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Scoliosis (HP:0002650)	Kyphosis [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Scoliosis (HP:0002650)	Scoliosis [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Seizures (HP:0001250)	Seizures [HS]	MIM:277590
Sotos syndrome (MIM:117550)	NSD1 (64324)	Tall stature (HP:0000098)	Overgrowth [HS]	MIM:130650
Sotos syndrome (MIM:117550)	PROP1 (5626)	Seizures (HP:0001250)	Seizures [HS]	MIM:262600
Sotos syndrome (MIM:117550)	PROP1 (5626)	Tall stature (HP:0000098)	Short_stature [HS]	MIM:262600
Sotos syndrome (MIM:117550)	SLC34A1 (6569)	Joint laxity (HP:0001388)	Increased_susceptibility_to_fractures [HS]	MIM:612286
Sotos syndrome (MIM:117550)	SNCB (6620)	Partial agenesis of the corpus callosum (HP:0001338)	abnormal_striatum_morphology [MP]	-
WAGR 11p13 deletion syndrome	PAX6 (5080)	Aniridia (HP:0000526)	iris_hypoplasia [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome	PAX6 (5080)	Aniridia (HP:0000526)	abnormally_decreased_size_eyelid [ZP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome	PAX6 (5080)	Aniridia (HP:0000526)	anterior_polar_cataracts [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_lens_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome	PAX6 (5080)	Aniridia (HP:0000526)	cataracts [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome	PAX6 (5080)	Aniridia (HP:0000526)	corneal_opacity [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_corneal_epithelium_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_iris_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome	PAX6 (5080)	Aniridia (HP:0000526)	Aniridia [HS, MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome	PAX6 (5080)	Aniridia (HP:0000526)	anophthalmia [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome	PAX6 (5080)	Aniridia (HP:0000526)	microphthalmia [MP]	PMID:16199712, MIM:148190

WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	delayed_eyelid_fusion [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	Optic_nerve_hypoplasia [HS, MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_pupil_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	anterior_iris_synechia [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	coloboma [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_lens_epithelium_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormally_disrupted_eye_development [ZP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormally_disrupted_lens_development_in_camera-type_eye [ZP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	absent_choroid Plexus [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormally_aplastic_lens [ZP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormally_mislocalised_cornea [ZP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormally_protruding_lens [ZP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormally_morphology_lens [ZP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormally_elliptic_lens [ZP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_anterior_stroma_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	Hereditary_childhood_corneal_clouding [HS]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	corneal_vascularization [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_lens_development [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	aphakia [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	Bilateral_coloboma_of_optic_nerve [HS]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_ciliary_body_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	corneal-lenticular_stalk [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormally_quality_lens [ZP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	White_corneal_opacification [HS]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_Bowman_membrane [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_anterior_eye_segment_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_iris_stroma_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_eye_anterior_chamber_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	fused_cornea_and_lens [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	small_lens [ZP, MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_lens_induction [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_lens Vesicle_development [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_iridocorneal_angle [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_eye_development [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	Presenile_cataracts [HS]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	mydriasis [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	irregularly_shaped_pupil [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	retina_hypoplasia [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_optic_cup_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	Irido-fundal_coloboma [HS]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	Foveal_hypoplasia [HS]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	absent_trabecular_meshwork [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	absent_Schlemm's_canal [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_optic_vesicleFormation [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_lens_fiber_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_cornea_endothelium_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_corneal_stroma_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	absent_lens_vesicle [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	Keratitis [HS]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Aniridia (HP:0000526)	abnormal_cornea_morphology [MP]	PMID:16199712, MIM:148190
WAGR 11p13 deletion syndrome (MIM:194072)	PAX6 (5080)	Intellectual disability (HP:0001249)	Intellectual_disability [HS]	MIM:206700
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Abnormality of the vagina (HP:0000142)	Primary_amenorrhea [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Cryptorchidism (HP:0000028)	Ambiguous_genitalia_male [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Cryptorchidism (HP:0000028)	Male_pseudohermaphroditism [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Cryptorchidism (HP:0000028)	abnormal_Sertoli_cell_development [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Cryptorchidism (HP:0000028)	abnormal_seminiferous_tubule_morphology [MP]	PMID:16199712

WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Cryptorchidism (HP:0000028)	abnormal_testis_morphology [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Cryptorchidism (HP:0000028)	Ambiguous_genitalia,_female [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Cryptorchidism (HP:0000028)	Gonadal_tissue_inappropriate_for_external_genitalia_or_chromosomal_abnormal_male_reproductive_system_morphology [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Cryptorchidism (HP:0000028)	abnormal_Leydig_cell_morphology [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Hypospadias (HP:0000047)	Ambiguous_genitalia,_male [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Hypospadias (HP:0000047)	Male_pseudohermaphroditism [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Hypospadias (HP:0000047)	Ambiguous_genitalia,_female [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Hypospadias (HP:0000047)	Gonadal_tissue_inappropriate_for_external_genitalia_or_chromosomal	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Nephroblastoma (Wilms tumor) (HP:0002667)	Ovarian_gonadoblastoma [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Nephroblastoma (Wilms tumor) (HP:0002667)	Nephroblastoma_(Wilms_tumor) [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Nephropathy (HP:0000112)	Nephropathy [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Renal failure (HP:0000083)	kidney_failure [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Renal failure (HP:0000083)	increased_urine_protein_level [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Renal failure (HP:0000083)	Nephrotic_syndrome [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Renal failure (HP:0000083)	glomerulonephritis [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Renal failure (HP:0000083)	chronic/endstage_renal_failure [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Renal failure (HP:0000083)	increased_kidney_apoptosis [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Streak ovary (HP:0010464)	True_hermaphroditism [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Streak ovary (HP:0010464)	primary_sex_reversal [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Streak ovary (HP:0010464)	Primary_amenorrhea [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Streak ovary (HP:0010464)	streak_gonad [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Streak ovary (HP:0010464)	Ambiguous_genitalia,_female [HS]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Streak ovary (HP:0010464)	absent_ovary [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Streak ovary (HP:0010464)	agonadal [MP]	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Streak ovary (HP:0010464)	Gonadal_tissue_inappropriate_for_external_genitalia_or_chromosomal	PMID:16199712
WAGR 11p13 deletion syndrome (MIM:194072)	WT1 (7490)	Streak ovary (HP:0010464)	testis_hypoplasia [MP]	PMID:16199712
Williams-Beuren syndrome (MIM:194050)	ABHD11 (83451)	Blue irides (HP:0000635)	abnormally_abolished_pigmentation [ZP]	-
Williams-Beuren syndrome (MIM:194050)	ABHD11 (83451)	Blue irides (HP:0000635)	abnormally_disrupted_pigmentation [ZP]	-
Williams-Beuren syndrome (MIM:194050)	ABHD11 (83451)	Premature graying of hair (HP:0002216)	abnormally_abolished_pigmentation [ZP]	-
Williams-Beuren syndrome (MIM:194050)	ABHD11 (83451)	Premature graying of hair (HP:0002216)	abnormally_disrupted_pigmentation [ZP]	-
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Coronary artery stenosis (HP:0005145)	aorta_coarctation [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Flattened nasal bridge (HP:0000425)	short_nasal_bone [MP]	-
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Hypercalcemia (HP:0003072)	hypercalcemia [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Hypodontia (HP:0000668)	abnormalFourth_branchial_arch_morphology [MP]	-
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Microdontia (HP:0000691)	abnormalFourth_branchial_arch_morphology [MP]	-
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Misalignment of teeth (HP:0000692)	malocclusion [MP]	-
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Misalignment of teeth (HP:0000692)	abnormalFourth_branchial_arch_morphology [MP]	-
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Nasal hypoplasia (HP:0003196)	short_snout [MP]	-
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	peripheral pulmonary artery stenosis (HP:0004969)	aorta_coarctation [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Pulmonic stenosis (HP:0001642)	increased_heart_right_ventricle_size [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Pulmonic stenosis (HP:0001642)	dilated_heart_right_ventricle [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Pulmonic stenosis (HP:0001642)	muscular_ventricular_septal_defect [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Supravalvular aortic stenosis (HP:0004381)	increased_heart_left_ventricle_size [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Supravalvular aortic stenosis (HP:0004381)	muscular_ventricular_septal_defect [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	BAZ1B (9031)	Supravalvular aortic stenosis (HP:0004381)	dilated_heart_left_ventricle [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	CLIP2 (7461)	Short stature (HP:0004322)	decreased_body_length [MP]	-
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Coronary artery stenosis (HP:0005145)	artery_stenosis [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Coronary artery stenosis (HP:0005145)	increased_aorta_wall_thickness [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Coronary artery stenosis (HP:0005145)	decreased_aorta_wall_thickness [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Coronary artery stenosis (HP:0005145)	abnormal_subclavian_artery_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Coronary artery stenosis (HP:0005145)	Peripheral_arterial_stenosis [HS]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Coronary artery stenosis (HP:0005145)	abnormal_artery_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Coronary artery stenosis (HP:0005145)	abnormal_artery_development [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Coronary artery stenosis (HP:0005145)	abnormal_aorta_elastic_tissue_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Coronary artery stenosis (HP:0005145)	decreased_aorta_elastin_content [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Coronary artery stenosis (HP:0005145)	abnormal_aorta_wall_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Cutis laxa (HP:0000973)	Loose_redundant_skin [HS]	-
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Mitral regurgitation (HP:0001653)	Aortic_insufficiency [HS]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Mitral regurgitation (HP:0001653)	Mitral_regurgitation [HS]	PMID:20089974

Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	artery_stenosis [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	increased_aorta_wall_thickness [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	decreased_aorta_wall_thickness [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	abnormal_subclavian_artery_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	abnormal_pulmonary_artery_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	Pulmonary_artery_stenosis [HS]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	Peripheral_arterial_stenosis [HS]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	abnormal_artery_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	abnormal_artery_development [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	abnormal_aorta_elastic_tissue_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	decreased_aorta_elastin_content [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	peripheral pulmonary artery stenosis (HP:0004969)	abnormal_aorta_wall_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Pulmonic stenosis (HP:0001642)	increased_aorta_wall_thickness [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Pulmonic stenosis (HP:0001642)	decreased_aorta_wall_thickness [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Pulmonic stenosis (HP:0001642)	abnormal_subclavian_artery_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Pulmonic stenosis (HP:0001642)	abnormal_artery_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Pulmonic stenosis (HP:0001642)	abnormal_artery_development [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Pulmonic stenosis (HP:0001642)	abnormal_aorta_elastic_tissue_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Pulmonic stenosis (HP:0001642)	decreased_aorta_elastin_content [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Pulmonic stenosis (HP:0001642)	abnormal_aorta_wall_morphology [MP]	PMID:20089974, MIM:185500
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Sloping shoulders (HP:0001556)	Inguinal_hernia [HS]	-
Williams-Beuren syndrome (MIM:194050)	ELN (2006)	Supravalvular aortic stenosis (HP:0004381)	Supravalvular_aortic_stenosis [HS]	MIM:185500
Williams-Beuren syndrome (MIM:194050)	FKBP6 (8468)	Urethral stenosis (HP:0008661)	testis_hypoplasia [MP]	-
Williams-Beuren syndrome (MIM:194050)	FZD9 (8326)	Joint laxity (HP:0001388)	abnormal_osteoblast_physiology [MP]	-
Williams-Beuren syndrome (MIM:194050)	FZD9 (8326)	Joint laxity (HP:0001388)	abnormal_skeleton_physiology [MP]	-
Williams-Beuren syndrome (MIM:194050)	FZD9 (8326)	Joint laxity (HP:0001388)	decreased_bone_strength [MP]	-
Williams-Beuren syndrome (MIM:194050)	FZD9 (8326)	Osteoporosis (HP:0000939)	decreased_bone_mineral_density [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	FZD9 (8326)	Reduced bone mineral density (HP:0004349)	decreased_bone_mineral_density [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	FZD9 (8326)	Short stature (HP:0004322)	decreased_body_length [MP]	PMID:20089974
Williams-Beuren syndrome (MIM:194050)	GTF2IRD1 (9569)	Anxiety (HP:0000739)	decreased_anxiety-related_response [MP]	PMID:10198167, PMID:16293761
Williams-Beuren syndrome (MIM:194050)	GTF2IRD1 (9569)	Anxiety (HP:0000739)	decreased_fear-related_response [MP]	PMID:10198167, PMID:16293761
Williams-Beuren syndrome (MIM:194050)	GTF2IRD1 (9569)	Anxiety (HP:0000739)	decreased_aggression_towards_mice [MP]	PMID:10198167, PMID:16293761
Williams-Beuren syndrome (MIM:194050)	GTF2IRD1 (9569)	Cutis laxa (HP:0000973)	palor [MP]	-
Williams-Beuren syndrome (MIM:194050)	GTF2IRD1 (9569)	Kyphoscoliosis (HP:0002751)	kyphosis [MP]	-
Williams-Beuren syndrome (MIM:194050)	GTF2IRD1 (9569)	Misalignment of teeth (HP:0000692)	malocclusion [MP]	-
Williams-Beuren syndrome (MIM:194050)	GTF2IRD1 (9569)	Nasal hypoplasia (HP:0003196)	short_snout [MP]	PMID:10198167, PMID:16293761
Williams-Beuren syndrome (MIM:194050)	GTF2IRD1 (9569)	Nasal hypoplasia (HP:0003196)	asymmetric_snout [MP]	PMID:10198167, PMID:16293761
Williams-Beuren syndrome (MIM:194050)	GTF2IRD1 (9569)	Premature graying of hair (HP:0002216)	belly_spot [MP]	-
Williams-Beuren syndrome (MIM:194050)	LIMK1 (3984)	Attention deficit hyperactivity disorder (HP:0007018)	hyperactivity [MP]	PMID:8689688, PMID:14556246, PMID:14745832
Williams-Beuren syndrome (MIM:194050)	MLXIPL (51085)	Diabetes mellitus (HP:0000819)	increased_liver_glycogen_level [MP]	-
Williams-Beuren syndrome (MIM:194050)	MLXIPL (51085)	Diabetes mellitus (HP:0000819)	abnormal_glucose_homeostasis [MP]	-
Williams-Beuren syndrome (MIM:194050)	MLXIPL (51085)	Diabetes mellitus (HP:0000819)	insulin_resistance [MP]	-
Williams-Beuren syndrome (MIM:194050)	MLXIPL (51085)	Diabetes mellitus (HP:0000819)	increased_circulating_glucose_level [MP]	-
Williams-Beuren syndrome (MIM:194050)	MLXIPL (51085)	Diabetes mellitus (HP:0000819)	increased_circulating_insulin_level [MP]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Joint laxity (HP:0001388)	Osteomyelitis_due_to_immunodeficiency [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Rectal prolapse (HP:0002035)	Rectal_abscess [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Rectal prolapse (HP:0002035)	Perirectal_abscesses_due_to_immunodeficiency [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent otitis media (HP:0000403)	Recurrent_Klebsiella_infections [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent otitis media (HP:0000403)	Recurrent_E_coli_infections [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent otitis media (HP:0000403)	Recurrent_Serratia_marcescens_infections [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent otitis media (HP:0000403)	Recurrent_bacterial_skin_infections [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent otitis media (HP:0000403)	Hepatic_abscesses_due_to_immunodeficiency [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent otitis media (HP:0000403)	Recurrent_Burkholderia_cepacia_infections [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent urinary tract infections (HP:0000010)	Recurrent_Klebsiella_infections [HS]	-

Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent urinary tract infections (HP:0000010)	Recurrent_E._coli_infections [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent urinary tract infections (HP:0000010)	Recurrent_Serratia_marcescens_infections [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent urinary tract infections (HP:0000010)	Recurrent_Staphylococcus_aureus_infections [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent urinary tract infections (HP:0000010)	Recurrent_Aspergillus_infections [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent urinary tract infections (HP:0000010)	Recurrent_bacterial_skin_infections [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent urinary tract infections (HP:0000010)	Hepatic_abscesses_due_to_immunodeficiency [HS]	-
Williams-Beuren syndrome (MIM:194050)	NCF1 (653361)	Recurrent urinary tract infections (HP:0000010)	Recurrent_Burkholderia_cepacia_infections [HS]	-
Williams-Beuren syndrome (MIM:194050)	STX1A (6804)	Anxiety (HP:0000739)	abnormal_response_to_novel_object [MP]	PMID:10874638 (regulatory region of STX1A deleted); PMID:22048961
Wolf-Hirschhorn syndrome (MIM:194190)	CTBP1 (1487)	Kyphosis (HP:0002808)	abnormal_spine_curvature [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	CTBP1 (1487)	Rib fusion (HP:0000902)	abnormal_rib_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	CTBP1 (1487)	Rib fusion (HP:0000902)	abnormal_diaphragm_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	CTBP1 (1487)	Rib segmentation abnormalities (HP:0006655)	abnormal_rib_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	CTBP1 (1487)	Rib segmentation abnormalities (HP:0006655)	abnormal_diaphragm_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	CTBP1 (1487)	Scoliosis (HP:0002650)	abnormal_spine_curvature [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	CTBP1 (1487)	Sternal ossification center abnormalities (HP:0006624)	abnormal_diaphragm_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	CTBP1 (1487)	Vertebral fusion (HP:0002948)	abnormal_embryonic_neuroepithelium_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	CTBP1 (1487)	Vertebral fusion (HP:0002948)	abnormal_neural_tube_closure [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Broad nasal bridge (HP:0000431)	Aplasia_of_the_parotid_gland [HS]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Broad nasal bridge (HP:0000431)	short_snout [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Broad nasal bridge (HP:0000431)	short_nasal_bone [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Broad nasal bridge (HP:0000431)	Malar_hypoplasia [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Broad nasal bridge (HP:0000431)	abnormal_snout_morphology [MP]	PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Broad nasal bridge (HP:0000431)	Low_nasal_bridge [HS]	PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Broad nasal bridge (HP:0000431)	abnormal_zygomatic_bone_morphology [MP]	PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Cleft lip/palate (HP:0000202)	abnormal_maxillary_shelf_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Cleft lip/palate (HP:0000202)	High-arched_palate [HS]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Conductive hearing impairment (HP:0000405)	abnormal_malleus_morphology [MP]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Conductive hearing impairment (HP:0000405)	abnormal_middle_ear_ossicle_morphology [MP]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Conductive hearing impairment (HP:0000405)	abnormal_stapes_morphology [MP]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Conductive hearing impairment (HP:0000405)	abnormal_incus_morphology [MP]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Conductive hearing impairment (HP:0000405)	impaired_hearing [MP]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Conductive hearing impairment (HP:0000405)	Mixed_hearing_loss [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Conductive hearing impairment (HP:0000405)	deafness [MP]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Conductive hearing impairment (HP:0000405)	decreased_brainstem_auditory_evoked_potential [MP]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Conductive hearing impairment (HP:0000405)	absent_brainstem_auditory_evoked_potential [MP]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Conductive hearing impairment (HP:0000405)	Recurrent_otitis_media_in_infancy [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Conductive hearing impairment (HP:0000405)	Recurrent_otitis_media_in_infancy_and_childhood [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Craniofacial asymmetry (HP:0004484)	Severe_cloverleaf_skull [HS]	MIM:602849, PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Craniofacial asymmetry (HP:0004484)	Coronal_craniosynostosis [HS]	MIM:602849, PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Craniofacial asymmetry (HP:0004484)	Brachycephaly [HS]	MIM:602849, PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Craniofacial asymmetry (HP:0004484)	Plagiocephaly [HS]	MIM:602849, PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Craniofacial asymmetry (HP:0004484)	domed_cranium [MP]	MIM:602849, PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Craniofacial asymmetry (HP:0004484)	Conical_incisor [HS]	MIM:602849, PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Cryptorchidism (HP:0000028)	Cryptorchidism [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Decreased fetal movement (HP:0001558)	Decreased_fetal_movement [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Epicanthus (HP:0000286)	Periorbital_fullness [HS]	MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Epicanthus (HP:0000286)	Telecanthus [HS]	MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Epicanthus (HP:0000286)	Downslanted_palpebral_fissures [HS]	MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Generalized hypotonia (HP:0001290)	Infantile_muscular_hypotonia [HS]	MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hemangiomas (HP:0001028)	Teratoma [HS]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hemangiomas (HP:0001028)	increased_lung_tumor_incidence [MP]	MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hemangiomas (HP:0001028)	skin_papilloma [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	High forehead (HP:0000348)	Broad_forehead [HS]	MIM:146000, MIM:602849, PMID:9321756, MIM:149730

Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	High forehead (HP:0000348)	Frontal_bossing [HS]	MIM:146000, MIM:602849, PMID:9321756, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Highly arched eyebrow (HP:0002553)	Periorbital_fullness [HS]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hyperconvex fingernails (HP:0001812)	2-3_finger_syndactyly [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hyperconvex fingernails (HP:0001812)	Small_thenar_eminence [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hyperconvex fingernails (HP:0001812)	Joint_contractures_involving_the_joints_of_the_hand [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hyperconvex fingernails (HP:0001812)	Trident_abnormality [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hypertelorism (HP:0000316)	Hypertelorism [HS, MP]	MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hypodontia (HP:0000668)	Delayed_eruption_of_primary_teeth [HS]	MIM:149730,-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hypodontia (HP:0000668)	malocclusion [MP]	MIM:149730,-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hypodontia (HP:0000668)	Hypodontia [HS]	MIM:149730,-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hypodontia (HP:0000668)	Carious_teeth [HS]	MIM:149730,-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hypodontia (HP:0000668)	Hypoplasia_of_dental_enamel [HS]	MIM:149730,-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hypodontia (HP:0000668)	abnormal_incisor_morphology [MP]	MIM:149730,-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hypodontia (HP:0000668)	long_incisors [MP]	MIM:149730,-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hypospadias (HP:0000047)	abnormal_urinary_bladder_urothelium_morphology [MP]	MIM:109800, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hypospadias (HP:0000047)	Coronal_hypospadias [HS]	MIM:109800, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Hypospadias (HP:0000047)	Transitional_cell_carcinoma_of_the_bladder [HS]	MIM:109800, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Kyphosis (HP:0002808)	kyphosis [MP]	PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Kyphosis (HP:0002808)	Lumbar_kyphosis_in_infancy [HS]	PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Kyphosis (HP:0002808)	abnormal_spine_curvature [MP]	PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Kyphosis (HP:0002808)	Lumbar_hyperlordosis [HS]	PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Low posterior hairline (HP:0002162)	Low_anterior_hairline [HS]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Microcephaly (HP:0000252)	Microcephaly [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Microcephaly (HP:0000252)	megacephaly [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Microcephaly (HP:0000252)	Megalencephaly [HS]	MIM:273300
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Micrognathia (HP:0000347)	Small_abnormally_formed_scapulae [HS]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Micrognathia (HP:0000347)	micromelia [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Micrognathia (HP:0000347)	short_vertebral_column [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Micrognathia (HP:0000347)	decreased_craniun_height [MP]	PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Micrognathia (HP:0000347)	Rhizomelic_shortening [HS]	PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Micrognathia (HP:0000347)	prognathia [MP]	PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Nystagmus (HP:0000639)	abnormal_malleus_morphology [MP]	MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Nystagmus (HP:0000639)	abnormal_middle_ear_ossicle_morphology [MP]	MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Nystagmus (HP:0000639)	abnormal_stapes_morphology [MP]	MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Nystagmus (HP:0000639)	abnormal_incus_morphology [MP]	MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Nystagmus (HP:0000639)	Recurrent_otitis_media_in_infancy [HS]	MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Nystagmus (HP:0000639)	Recurrent_otitis_media_in_infancy_and_childhood [HS]	MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Poorly formed pinnae (HP:0008562)	Cupped_ear [HS]	MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Prominent glabella (HP:0002057)	Frontal_bossing [HS]	PMID:9321756
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	Short_middle_phalanges_(feet) [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	2-3_finger_syndactyly [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	Small_thenar_eminence [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	Hypoplasia_of_the_middle_phalanges_of_the_hand [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	Partial_duplication_of_the_phalanges_of_the_thumb [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	Bilateral_triphalangeal_thumbs [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	Joint_contractures_involving_the_joints_of_the_hand [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	Trident_abnormality [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	Broad_thimble-like_middle_phalanges [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	Clindodactyly,_3.5_finger [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	Anopla_of_the_proximal_phalanx_of_the_thumb [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Pseudoepiphyses of the metacarpals (HP:0009193)	Cone-shaped_epiphyses_of_the_phalanges_of_the_hand [HS]	MIM:146000,
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Ptosis (HP:0000508)	Periorbital_fullness [HS]	MIM:602849, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Ptosis (HP:0000508)	Ptosis [HS]	MIM:602849, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Ptosis (HP:0000508)	Telecanthus [HS]	MIM:602849, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Ptosis (HP:0000508)	Downslanted_palpebral_fissures [HS]	MIM:602849, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_tibia_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_femur_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_fibula_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	increased_diameter_of_long_bones [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	increased_diameter_of_femur [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	bowed_fibula [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_long_bone_morphology [MP]	MIM:187600

Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_costal_cartilage_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_long_bone_metaphysis_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	Marked_shortness_and_bowing_of_long_bones [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	short_tubular_bones_with_mild_metaphyseal_flare [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	long_radius [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_long_bone_hypertrophic_chondrocyte_zone [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	Broad_femoral_metaphyses [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	Short_and_small_iliac_bones [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_long_bone_epiphyseal_plate_proliferative_zone [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	short_femur [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_ulna_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	Hypoplasia_of_the_radius [HS, MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	bowed_radius [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	bowed_femur [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	bowed_tibia [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	short_tibia [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	Narrow_chest [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_rib_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	Short_ribs [HS, MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	Genu_varum [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	increased_long_bone_epiphyseal_plate_size [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	disorganized_long_bone_epiphyseal_plate [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	decreased_long_bone_epiphyseal_plate_size [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_long_bone_epiphyseal_plate_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_vertebral_epiphyseal_plate_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_thoracic_cage_shape [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	small_thoracic_cage [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_thoracic_vertebrae_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_long_bone_diaphysis_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	Hypoplasia_of_the_ulna [HS, MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	short_humerus [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	long_ulna [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	decreased_length_of_long_bones [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	small_thoracic_cavity [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	bowed_ulna [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	Flared_irregular_metaphyses [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	decreased_width_of_hypertrophic_chondrocyte_zone [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	increased_width_of_hypertrophic_chondrocyte_zone [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	Wide-cupped_costochondral_junctions [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	long_tibia [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	long_humerus [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	abnormal_humerus_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	long_femur [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib fusion (HP:0000902)	bowed_humerus [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_tibia_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_femur_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_fibula_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	increased_diameter_of_long_bones [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	increased_diameter_of_femur [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	bowed_fibula [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_long_bone_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_costal_cartilage_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_long_bone_metaphysis_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	Marked_shortness_and_bowing_of_long_bones [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	short_tubular_bones_with_mild_metaphyseal_flare [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	long_radius [MP]	MIM:187600

Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_long_bone_hypertrophic_chondrocyte_zone [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	Broad_femoral_metaphyses [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	Short_and_small_iliac_bones [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_long_bone_epiphyseal_plate_proliferative_zone [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	short_femur [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_ulna_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	Hypoplasia_of_the_radius [HS, MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	bowed_radius [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	bowed_femur [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	bowed_tibia [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	short_tibia [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	Narrow_chest [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_rib_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	Short_ribs [HS, MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	Genu_varum [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	increased_long_bone_epiphyseal_plate_size [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	disorganized_long_bone_epiphyseal_plate [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	decreased_long_bone_epiphyseal_plate_size [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_long_bone_epiphyseal_plate_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_vertebral_epiphyseal_plate_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_thoracic_cage_shape [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	small_thoracic_cage [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_thoracic_vertebrae_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_long_bone_diaphysis_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	Hypoplasia_of_the_ulna [HS, MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	short_humerus [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	long_ulna [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	decreased_length_of_long_bones [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	small_thoracic_cavity [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	bowed_ulna [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	Flared_irregular_metaphyses [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	decreased_width_of_hypertrophic_chondrocyte_zone [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	increased_width_of_hypertrophic_chondrocyte_zone [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	Wide-cupped_costochondral_junctions [HS]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	long_tibia [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	long_humerus [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	abnormal_humerus_morphology [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	long_femur [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rib segmentation abnormalities (HP:0006655)	bowed_humerus [MP]	MIM:187600
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Rieger anomaly (HP:0000558)	Recurrent_corneal_erusions [HS]	MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Scoliosis (HP:0002650)	kyphoscoliosis [MP]	PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Scoliosis (HP:0002650)	Scoliosis [HS, MP]	PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Scoliosis (HP:0002650)	abnormal_spine_curvature [MP]	PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Scoliosis (HP:0002650)	Lumbar_hyperlordosis [HS]	PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	increased_cochlear_outer_hair_cell_number [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	abnormal_pillar_cell_morphology [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	abnormal_cochlear_outer_hair_cell_morphology [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	abnormal_cochlea_morphology [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	abnormal_organ_of_Corti_morphology [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	abnormal_organ_of_Corti_supporting_cell_differentiation [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	abnormal_pillar_cell_differentiation [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	absent_tunnel_of_Corti [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	abnormal_patterning_of_the_organ_of_Corti [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	abnormal_cochlear_OHC_efferent_innervation_pattern [MP]	MIM:602849

Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	impaired_hearing [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	sensorineural_hearing_loss [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	Sensorineural_hearing_impairment [HS]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	deafness [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	decreased_brainstem_auditory_evoked_potential [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	absent_brainstem_auditory_evoked_potential [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	increased_Deiters_cell_number [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	abnormal_Deiters_cell_morphology [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sensorineural hearing impairment (HP:0000407)	absent_pilar_cells [MP]	MIM:602849
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Short stature (HP:0004322)	Tall_stature [HS]	MIM:610474, MIM:187600, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Short stature (HP:0004322)	Neonatal_short-limbed_dwarfism [HS]	MIM:610474, MIM:187600, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Short stature (HP:0004322)	Short-limb_dwarfism_identifiable_during_childhood [HS]	MIM:610474, MIM:187600, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Short stature (HP:0004322)	Lethal_micromelic_dwarfism [HS]	MIM:610474, MIM:187600, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Short stature (HP:0004322)	decreased_body_length [MP]	MIM:610474, MIM:187600, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sternal ossification center abnormalities (HP:0006624)	abnormal_sternum_ossification [MP]	MIM:187600, PMID:9321756, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sternal ossification center abnormalities (HP:0006624)	Pectus_excavatum [HS]	MIM:187600, PMID:9321756, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sternal ossification center abnormalities (HP:0006624)	Absent_ossification/absence_of_radius [HS]	MIM:187600, PMID:9321756, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sternal ossification center abnormalities (HP:0006624)	small_thoracic_cavity [MP]	MIM:187600, PMID:9321756, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sternal ossification center abnormalities (HP:0006624)	abnormal_sternebra_morphology [MP]	MIM:187600, PMID:9321756, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Sternal ossification center abnormalities (HP:0006624)	decreased_bone_mineral_density [MP]	MIM:187600, PMID:9321756, MIM:149730
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Talipes (HP:0001883)	Small_thenar_eminence [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Talipes (HP:0001883)	Small_sacroiliac_notches [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Talipes (HP:0001883)	Limited_elbow_and_hip_extension [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Talipes (HP:0001883)	Joint_contractures_involving_the_joints_of_the_hand [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Talipes (HP:0001883)	Camptodactyly_(feet) [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Talipes (HP:0001883)	Short_femoral_neck [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Talipes (HP:0001883)	Broad_hallux [HS]	MIM:149730, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Vertebral fusion (HP:0002948)	Capitate-hamate_fusion [HS]	MIM:602849, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Vertebral fusion (HP:0002948)	small_vertebrae [MP]	MIM:602849, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Vertebral fusion (HP:0002948)	Severe_platyspondyly [HS]	MIM:602849, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Vertebral fusion (HP:0002948)	small_caudal_vertebrae [MP]	MIM:602849, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Vertebral fusion (HP:0002948)	enlarged_vertebral_body [MP]	MIM:602849, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Vertebral fusion (HP:0002948)	short_vertebral_body [MP]	MIM:602849, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Vertebral fusion (HP:0002948)	elongated_vertebral_body [MP]	MIM:602849, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Vertebral fusion (HP:0002948)	abnormal_cranial_suture_morphology [MP]	MIM:602849, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Vertebral fusion (HP:0002948)	abnormal_sagittal_suture_morphology [MP]	MIM:602849, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Vertebral fusion (HP:0002948)	abnormal_coronal_suture_morphology [MP]	MIM:602849, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFR3 (2261)	Vertebral fusion (HP:0002948)	abnormal_lambdoidal_suture_morphology [MP]	MIM:602849, PMID:9321756, MIM:100800
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Craniofacial asymmetry (HP:0004484)	domed_cranium [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Gastroesophageal reflux (HP:0002020)	abnormal_cervical_vertebrae_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Gastroesophageal reflux (HP:0002020)	abnormal_cervical_atlas_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Intrauterine growth restriction (HP:0001511)	fetal_growth_retardation [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Iris coloboma (HP:0000612)	abnormally_disrupted_embryonic_camera-type_eye_development [ZP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Low posterior hairline (HP:0002162)	abnormal_cervical_vertebrae_morphology [MP]	-

Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Low posterior hairline (HP:0002162)	abnormal_cervical_atlas_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Micrognathia (HP:0000347)	axial_skeleton_hypoplasia [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Micrognathia (HP:0000347)	mandible_hypoplasia [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Micrognathia (HP:0000347)	abnormally_decreased_size_mandibular_arch_skeleton [ZP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Preauricular pit (HP:0004467)	abnormal_cervical_vertebrae_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Preauricular pit (HP:0004467)	abnormal_cervical_atlas_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib fusion (HP:0000902)	abnormal_pelvic_girdle_bone_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib fusion (HP:0000902)	abnormal_appendicular_skeleton_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib fusion (HP:0000902)	herniated_diaphragm [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib fusion (HP:0000902)	decreased_long_bone_epiphyseal_plate_size [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib fusion (HP:0000902)	abnormal_thyroid_cartilage_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib fusion (HP:0000902)	small_thoracic_cage [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib fusion (HP:0000902)	abnormal_cricoid_cartilage_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib fusion (HP:0000902)	thin_diaphragm_muscle [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib fusion (HP:0000902)	abnormal_diaphragm_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib segmentation abnormalities (HP:0006655)	small_thoracic_cavity [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib segmentation abnormalities (HP:0006655)	abnormal_pelvic_girdle_bone_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib segmentation abnormalities (HP:0006655)	abnormal_appendicular_skeleton_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib segmentation abnormalities (HP:0006655)	herniated_diaphragm [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib segmentation abnormalities (HP:0006655)	decreased_long_bone_epiphyseal_plate_size [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib segmentation abnormalities (HP:0006655)	abnormal_thyroid_cartilage_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib segmentation abnormalities (HP:0006655)	small_thoracic_cage [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib segmentation abnormalities (HP:0006655)	abnormal_cricoid_cartilage_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib segmentation abnormalities (HP:0006655)	thin_diaphragm_muscle [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib segmentation abnormalities (HP:0006655)	abnormal_diaphragm_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Rib segmentation abnormalities (HP:0006655)	small_thoracic_cavity [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Short stature (HP:0004322)	decreased_body_height [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Sternal ossification center abnormalities (HP:0006624)	abnormal_sternum_ossification [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Sternal ossification center abnormalities (HP:0006624)	abnormal_sternum_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Sternal ossification center abnormalities (HP:0006624)	herniated_diaphragm [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Sternal ossification center abnormalities (HP:0006624)	thin_diaphragm_muscle [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Sternal ossification center abnormalities (HP:0006624)	short_sternum [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Sternal ossification center abnormalities (HP:0006624)	abnormal_diaphragm_morphology [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Sternal ossification center abnormalities (HP:0006624)	small_thoracic_cavity [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Ventricular septal defect (HP:0001629)	pulmonary_valve_stenosis [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Ventricular septal defect (HP:0001629)	perimembranous_ventricular_septal_defect [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	FGFRL1 (53834)	Ventricular septal defect (HP:0001629)	muscular_ventricular_septal_defect [MP]	PMID:19383940
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Broad nasal bridge (HP:0000431)	abnormal_snout_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Broad nasal bridge (HP:0000431)	short_snout [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Broad nasal bridge (HP:0000431)	abnormal_zygomatic_bone_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Conductive hearing impairment (HP:0000405)	deafness [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Conductive hearing impairment (HP:0000405)	decreased_brainstem_auditory_evoked_potential [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Conductive hearing impairment (HP:0000405)	increased_susceptibility_to_otitis_media [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Gastroesophageal reflux (HP:0002020)	abnormal_cervical_vertebrae_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Hyperconvex fingernails (HP:0001812)	abnormal_autopod_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Hyperconvex fingernails (HP:0001812)	abnormal_digit_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Hypertelorism (HP:0000316)	enophthalmos [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Hypodontia (HP:0000668)	abnormal_incisor_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Kyphosis (HP:0002808)	kyphosis [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Low posterior hairline (HP:0002162)	sparse_hair [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Low posterior hairline (HP:0002162)	abnormal_cervical_vertebrae_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Low posterior hairline (HP:0002162)	disheveled_coat [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Low posterior hairline (HP:0002162)	rough_coat [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Microcephaly (HP:0000252)	broad_head [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Micrognathia (HP:0000347)	abnormal_mandibular_condyloid_process_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Nystagmus (HP:0000639)	decreased_startle_reflex [MP]	-

Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Nystagmus (HP:0000639)	increased_susceptibility_to_otitis_media [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Poorly formed pinnae (HP:0008562)	small_ears [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Praearicular pit (HP:0004467)	abnormal_cervical_vertebrae_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Proptosis (HP:0000520)	enophthalmos [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Pseudoepiphyses of the metacarpals (HP:0009193)	abnormal_autopod_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Pseudoepiphyses of the metacarpals (HP:0009193)	abnormal_digit_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib fusion (HP:0000902)	abnormal_tibia_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib fusion (HP:0000902)	increased_diameter_of_femur [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib fusion (HP:0000902)	abnormal_rib_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib fusion (HP:0000902)	abnormal_long_bone_diaphysis_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib fusion (HP:0000902)	abnormal_long_bone_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib fusion (HP:0000902)	abnormal_long_bone_hypertrophic_chondrocyte_zone [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib fusion (HP:0000902)	short_femur [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib segmentation abnormalities (HP:0006655)	abnormal_tibia_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib segmentation abnormalities (HP:0006655)	increased_diameter_of_femur [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib segmentation abnormalities (HP:0006655)	abnormal_rib_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib segmentation abnormalities (HP:0006655)	abnormal_long_bone_diaphysis_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib segmentation abnormalities (HP:0006655)	abnormal_long_bone_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib segmentation abnormalities (HP:0006655)	abnormal_long_bone_hypertrophic_chondrocyte_zone [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib segmentation abnormalities (HP:0006655)	short_femur [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Rib segmentation abnormalities (HP:0006655)	abnormal_tibia_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Sensorineural hearing impairment (HP:0000407)	spiral_ligament_degeneration [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Sensorineural hearing impairment (HP:0000407)	abnormal_inner_ear_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Sensorineural hearing impairment (HP:0000407)	organ_of_Corti_degeneration [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Sensorineural hearing impairment (HP:0000407)	cochlear_hair_cell_degeneration [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Sensorineural hearing impairment (HP:0000407)	sensorineural_hearing_loss [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Sensorineural hearing impairment (HP:0000407)	deafness [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Sensorineural hearing impairment (HP:0000407)	decreased_brainstem_auditory_evoked_potential [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Stenotic external auditory canal (HP:0000402)	abnormal_external_auditory_canal_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Sternal ossification center abnormalities (HP:0006624)	osteosclerosis [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Talipes (HP:0001883)	abnormal_autopod_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Ventricular septal defect (HP:0001629)	abnormal_aortic_valve_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Ventricular septal defect (HP:0001629)	thick_ventricular_wall [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Ventricular septal defect (HP:0001629)	thick_interventricular_septum [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Ventricular septal defect (HP:0001629)	abnormal_heart_left_ventricle_outflow_tract_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Ventricular septal defect (HP:0001629)	increased_left_ventricle_weight [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Ventricular septal defect (HP:0001629)	abnormal_heart_ventricle_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	IDUA (3425)	Vertebral fusion (HP:0002948)	abnormal_joint_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	PDE6B (5158)	Iris coloboma (HP:0000612)	abnormal_eye_development [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	PDE6B (5158)	Nystagmus (HP:0000639)	impaired_pupillary_reflex [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	PDE6B (5158)	Poorly formed pinnae (HP:0008562)	abnormal_ear_pigmentation [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	PDE6B (5158)	Stenotic external auditory canal (HP:0000402)	abnormal_ear_pigmentation [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	TACC3 (10460)	Hemangiomas (HP:0001028)	T_cell_derived_lymphoma [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	TACC3 (10460)	Intrauterine growth restriction (HP:0001511)	embryonic_growth_retardation [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	TACC3 (10460)	Short stature (HP:0004322)	decreased_body_length [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	TACC3 (10460)	Vertebral fusion (HP:0002948)	lumbar_vertebral_fusion [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	TACC3 (10460)	Vertebral fusion (HP:0002948)	abnormal_vertebral_arch_morphology [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	WHSC1 (7468)	Atrial septal defect (HP:0001631)	abnormal_interatrial_septum_morphology [MP]	PMID:19483677
Wolf-Hirschhorn syndrome (MIM:194190)	WHSC1 (7468)	Atrial septal defect (HP:0001631)	atrial_septal_defect [MP]	PMID:19483677
Wolf-Hirschhorn syndrome (MIM:194190)	WHSC1 (7468)	Cleft lip/palate (HP:0000202)	cleft_palate [MP]	PMID:12563561
Wolf-Hirschhorn syndrome (MIM:194190)	WHSC1 (7468)	Hypodontia (HP:0000668)	malocclusion [MP]	PMID:12563561
Wolf-Hirschhorn syndrome (MIM:194190)	WHSC1 (7468)	Sternal ossification center abnormalities (HP:0006624)	failure_of_sternum_ossification [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	WHSC1 (7468)	Sternal ossification center abnormalities (HP:0006624)	abnormal_sternum_ossification [MP]	-
Wolf-Hirschhorn syndrome (MIM:194190)	WHSC1 (7468)	Ventricular septal defect (HP:0001629)	perimembranous_ventricular_septal_defect [MP]	PMID:19483677

Phenotypic overlap in the contribution of individual genes to CNV pathogenicity revealed by cross-species computational analysis of single-gene mutations in humans, mice and zebrafish:

Supplementary File S3

Doelken SC, Sebastian Köhler S, Mungall CJ, et al.

January 16, 2013

Contents

S3 Detailed Computational Methods	2
S3.1 Input Ontologies	3
S3.2 Uberon bridge to species anatomy ontologies	3
S3.3 Logical Definitions	4
S3.4 Generation of HPO and MPO alignments	5
S3.5 Compilation of annotation sets	6
S3.5.1 Generation of zebrafish phenotype ontology	6
S3.6 Construction of the “UberPheno” composite analysis ontology	7
S3.7 Mapping model organism phenotypes to human genes	7
S3.8 Orthologs	8
S3.9 P-value correlation	8

S3 Detailed Computational Methods

This section provides a detailed description of the methods used to generate the results in the main manuscript. A makefile that downloads all datasets and generates the *uberpheno* is available from the HPO-svn repository.¹ All executable jar files are stored there. A graphical illustration of the workflow and dataflow is shown in Figure S3.1.

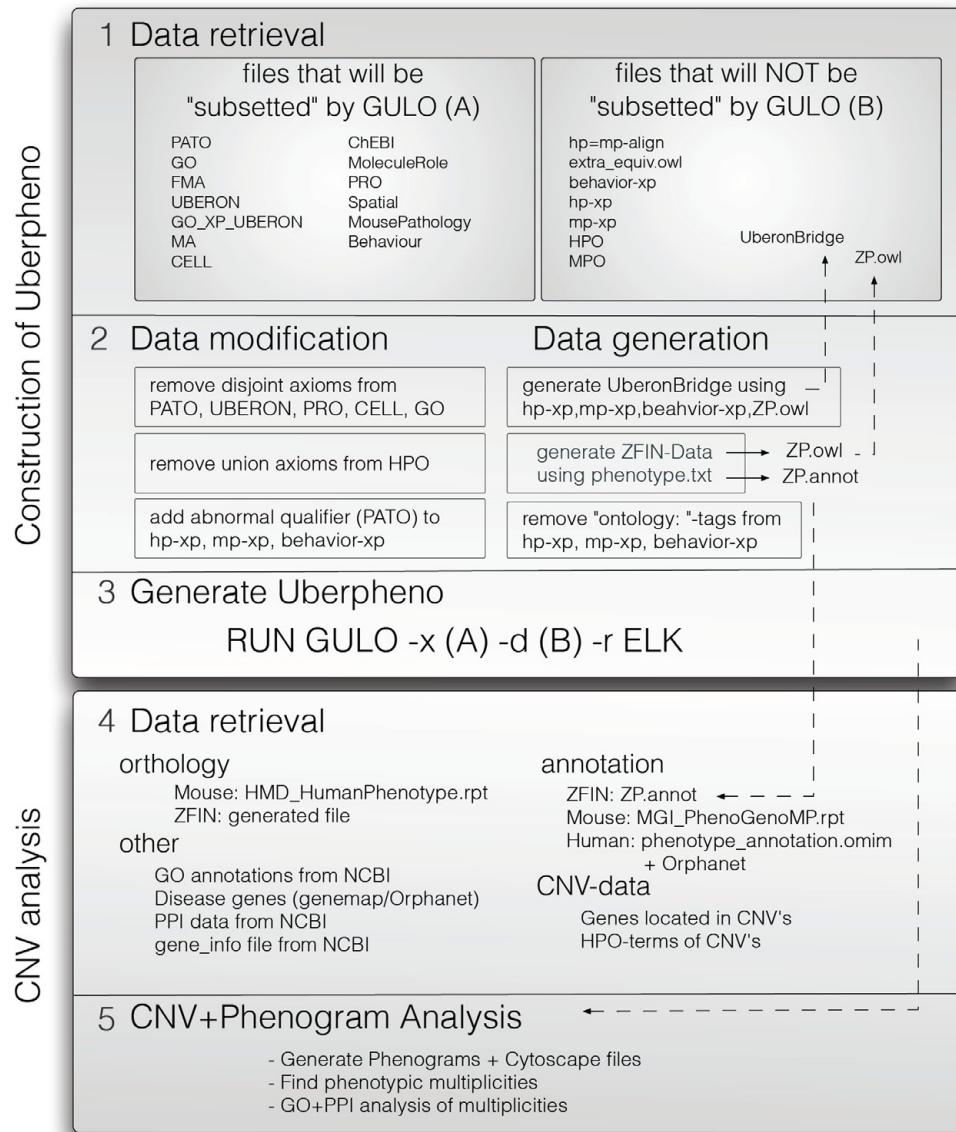


Figure S3.1 The work- and dataflow for generating the UberPheno ontology. It is also shown how the CNV analysis is performed.

¹<https://compbio.charite.de/svn/hpo/trunk/misc/uberpheno>

S3.1 Input Ontologies

The ontologies shown in Table were used to generate the composite ontology as described in section . The ontologies were obtained from the OBO Foundry homepage.² For the analysis described in the main manuscript, all files were downloaded on February 15, 2012.

Ontology	Description	File
IMR	Molecule role (INOH Protein name/family name ontology)	MoleculeRoleOntology.obo
MA	Mouse adult gross anatomy ¹	adult_mouse_anatomy.obo
BFO	Basic Formal Ontology ²	bfo-1.1.owl
CL	Cell Ontology ³	cell.obo
CHEBI	Chemical Entities of Biological Interest ⁴	chebi.obo
GO	Gene Ontology ⁵	gene_ontology.1_2.obo ³
HP	Human Phenotype Ontology ⁶	human-phenotype-ontology.obo
FMA	Foundational Model of Anatomy (adult human) ⁷	fma2_obo.obo
MP	Mammalian Phenotype Ontology ⁸	mammalian_phenotype.obo
NBO	Neuro Behavior Ontology	behavior.obo ⁴
MPATH	Mouse Pathology ⁹	mouse_pathology.obo
PR	Protein Ontology ¹⁰	pro.obo
PATO	Phenotypic Qualities ¹¹	quality.obo
BSPO	Spatial Ontology	spatial.obo
UBERON	Multi-species anatomy ¹²	uberon.obo
ZFA	Zebrafish anatomy and development ¹³	zebrafish_anatomy.obo

We use the OWL translation of all OBO-Format ontologies, using the OBO-Format to OWL conversion library (available from <http://code.google.com/p/oboformat>). Disjointedness axioms were removed from PATO, UBERON, PR and CL.

S3.2 Uberon bridge to species anatomy ontologies

Uberon is a multi-species anatomical ontology, and contains classes that are generalization of classes in ontologies such as MA, FMA and ZFA. For our analysis, this type of subclass relation was treated as an equivalentTo relation.¹² In order to construct UberPheno (See section), an equivalence axiom was generated for every class in Uberon U that contains a cross-reference to a species anatomy ontology class A :

`EquivalentClasses(U A)`

We created a Java program for generating this ontology, which can be executed as follows:

²<http://www.obofoundry.org/>

```

GenerateUberonBridge
-xp "equivalenceFiles/mp-equivalence-axioms.mod.obo,
    equivalenceFiles/hp-equivalence-axioms.mod.obo,
    behavior_xp.obo,
    zp.owl"
-uberon "data/uberon.obo"
-outfile "uberonbridge/uberphenoUberonbridge.owl"
-addAsEquivalence -excludeUpperLevel

```

The program can be downloaded from the project code archive.⁵ Note that we did not generate bridging axioms for UBERON classes that belong to the subset “upper_level”.

S3.3 Logical Definitions

Logical definitions have been developed for GO¹⁴, MPO¹⁵, and HPO (this work) that reference other ontologies. These bridging ontologies (also called cross-product files) are available on the main OBO Foundry website from <http://www.obofoundry.org/index.cgi?show=mappings>, as well as from the individual repositories for each of the projects.

Ontology	File	Size
HPO logical definitions ¹⁵	hp-equivalence-axioms.obo ⁶	4874
MPO logical definitions ¹⁵	mp-equivalence-axioms.obo ⁷	6679
GO logical definitions using Uberon ¹⁴	biological_process_xp_uber_anatomy.obo	1484
Behavior xp	behavior_xp.obo ⁸	110

We also created an additional ontology called `extra_equiv.owl` for use with the GO logical definitions.

The phenotype ontology logical definitions provide axioms that connect phenotype classes to multiple classes in most of the ontologies in the above table. An example (in OWL Manchester Syntax¹⁶) is:

Class: HP_0001650

EquivalentTo: PATO_0001847 and inheres_in some FMA_7236

⁵<https://compbio.charite.de/svn/hpo/trunk/misc/uberpheno>

Here `HP_0001650` is the URI for “aortic stenosis”, `PATO_0001847` is the URI for “constricted” and `FMA_7236` is the URI for “Aortic valve”. Logical definitions such as this connect the phenotype ontologies to a variety of other ontologies. A total of 5141 such EQ definitions were created for HPO terms and used for this project.

S3.4 Generation of HPO and MPO alignments

The HPO and MPO logical definitions were augmented with pairwise equivalence axioms generated by lexical matching. We generated the file `mp_hp-align-equiv.owl` (c.f. the project code archive⁹ or from the phenotype ontologies archive on Google code¹⁰).

An equivalence pair ($C1, C2$) is generated if $C1$ and $C2$ are reciprocal best matches according to names/synonyms, as follows.

Lexical matching: We use the SWI-Prolog porter-stem method in the `nlp` library¹⁷ to tokenize and stem all tokens for each synonym, omitting stemming for acronyms or special tokens (any token with >1 upper case character, or any non-alphanumeric characters). We filter out prepositions (e.g. “of”, “to”) and determinants (e.g. “a”, “the”). We apply special purpose substitutions (e.g. changing I, first, 1st to 1), and use the Obol library of biological relational adjectives¹⁸ to substitute adjectives such as “renal” with their noun form (e.g. “kidney”).

Finally, the normalized tokens are ordered alphabetically and concatenated without whitespace to produce a normalized label.

Reciprocal best matches: Any class pair matches if they share the same normalized label. A class pair $C1, C2$ is a best match if there is no $C2'$ such that $C1$ matches $C2'$, and the both synonyms used for the latter had stronger scope than both the synonyms used for the former. We treat exact synonyms and primary labels as equal to one another as well as stronger than broad, narrow or related synonyms. We only consider inter-ontology matches.

A best match $C1, C2$ is reciprocal if $C2$ has best match $C1$.

Each equivalence pair is then translated to OWL equivalence axioms:

`EquivalentClasses(C1 C2)`

We then checked all equivalence pairs for structural difference across the two ontologies, and resolved these.

Filtering: We maintain a manually edited file of explicitly disjoint MP-HP class pairs. The purpose of this file is to exclude false positive alignments based on non-exact synonyms. Any potential equivalence pair $C1 - C2$ is

⁹<https://compbio.charite.de/svn/hpo/trunk/misc/uberpheno>
¹⁰<http://code.google.com/p/phenotype-ontologies>

excluded if there is a disjoint pair $D_1 - D_2$ and C_1 and C_2 are inferred subclasses of D_1 and D_2 respectively (recall that subclass is a reflexive relation, so this includes the case where $C_1 = D_1$ and $C_2 = D_2$).

A total of 1064 such lexically derived equivalence axioms were derived in this way and used to supplement the semantic analysis.

S3.5 Compilation of annotation sets

The HPO⁶ has been used to annotate 5035 diseases listed in OMIM¹⁹. Similarly, the MPO has been used to annotate genetically modified mice at MGI²⁰, and (as described in Section), E/Q annotations have been used to describe the phenotypes of genetically modified zebrafish. A phenotypic annotation is a statement that a given disease is characterized by a phenotypic feature. Phenotypic annotations were downloaded from ZFIN,¹¹ MGI,¹² and the HPO.¹³ Additionally annotations for the 27 CNV diseases analyzed in this work were created by manual curation using Phenote.¹⁴ Information on the sources for annotation is given in the Supplementary Tables S4.

Annotations	Source
MGI mouse gene phenotype	MGI_PhenoGeno.rpt
ZFIN zebrafish gene phenotype	pheno_obo.txt
Human disorder phenotype	HPO annotation files

S3.5.1 Generation of zebrafish phenotype ontology

The ZFIN annotations are post-composed, using a combination of terms in an Entity-Quality model. We implemented the translation table described in ref.¹⁵ to generate the ontology `zp.owl`, creating classes in the ZP identifier space using the ZFIN annotation file as a source of distinct phenotypes.

For example, a zebrafish gene annotation with `Entity=ZFA:0000014` (dorsal aorta), `Quality=PATO:0013789` (decreased width) and `Qualifier=PATO:0000460` (abnormal) generates a class:

Class: ZP_0013789

Annotations: label "abnormally decreased width dorsal aorta"

EquivalentClassOf: PATO_0000599 and

¹¹http://zfin.org/zf_info/downloads.html

¹²<ftp://ftp.informatics.jax.org/pub/reports/index.html>

¹³<http://www.human-phenotype-ontology.org>

¹⁴<http://phenote.org>

```
inheres_in some ZFA_0000014 and
qualifier some PATO_0000460
```

S3.6 Construction of the “UberPheno” composite analysis ontology

We generated an ontology called “UberPheno” from the set of ontologies and bridging axioms above.

We first combine all ontologies and bridging axioms together into a single OWL ontology. We use the ELK reasoner²¹ to calculate subclass and equivalence relationships between classes. These steps are implemented within the GULO framework²², and can be recreated using the following command:

```
java -Xmx8G -jar jars/GULO.jar
-x "ls -m data/dataForUberpheno/subsettable_ontologies/*"
-d "ls -m data/dataForUberpheno/not_subsettable_ontologies/*"
-o data/uberphenoOut/
-r elk
```

We then use The Ontologizer API²³ to merge all clusters of equivalent classes together into a single class. The HPO identifier is taken as the primary identifier.

S3.7 Mapping model organism phenotypes to human genes

The file `biodata/MGIdata/HMD_HumanPhenotype.rpt` was used to determine the orthology relations between mouse and human genes²⁴. The file `biodata/MGIdata/MGI_PhenoGenoMP.rpt` was used to obtain mouse phenotype annotations²⁵. Note that annotations were included only if they applied to a gene with an identified human ortholog.

The file `data/ortho_gene.txt` was used to determine the orthology relations between zebrafish and human genes. The file `zp.annot` (generate by the `make-command`) was used to obtain zebrafish phenotype annotations²⁶.

Ontology and annotation files for human Mendelian disease were downloaded from the HPO Website²⁷. The `genemap` file from OMIM was used to associate genes with diseases¹⁹. Positional information for the human genes was obtained from NCBI Entrez Gene²⁸.

S3.8 Orthologs

There were 6535 mouse genes with phenotype information for which a human ortholog could be identified using information from the Mouse Genome Informatics (MGI)²⁰ sequence group,¹⁵ and there were 1625 zebrafish genes.¹⁶ In addition, there were 1843 human genes with phenotype information on monogenic diseases in the HPO. In all, there were 7546 human genes with phenotypic information either in human or one of the model organisms.

S3.9 P-value correlation

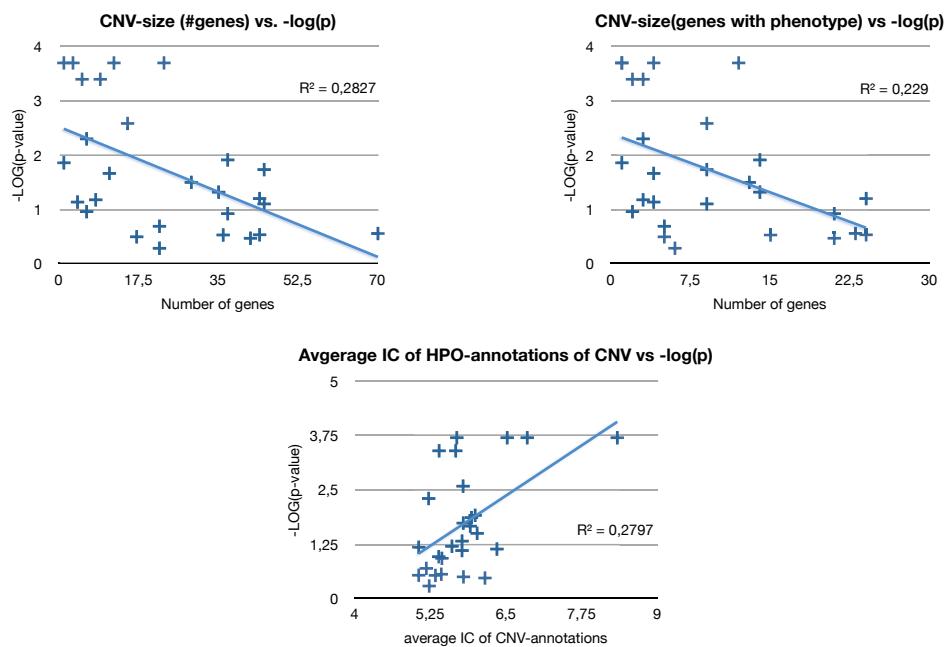


Figure S3.2 The P-values of the phenogram scores correlate with the size of the intervals measured by the absolute numbers of genes (**upper left**) and the numbers of genes with available phenotype information (**upper right**). Thus, a higher number of genes in a CNV interval is associated with a lesser degree of statistical significance of the phenogram. The P-values also correlate with the specificity of the phenotype information of the CNV disorders measured by the average information content (IC) of the CNV phenotypes (**bottom plot**). Thus unspecific phenotypic annotations (terms with low IC) are associated with less significant phenogram scores.

¹⁵<ftp://ftp.informatics.jax.org/pub/reports/index.html>

¹⁶http://zfin.org/zf_info/downloads.html

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Phenotypic overlap in the contribution of individual genes to CNV pathogenicity revealed by cross-species computational analysis of single-gene mutations in humans, mice and zebrafish:
Supplementary File S4

Doelken SC, Sebastian Köhler S, Mungall CJ, et al.

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Contents

S4 Annotations of CNV disorders	3
S4.1 Miller Dieker Lissencephaly Syndrome [MIM:247200]	4
S4.2 Williams Beuren Syndrome [MIM:194050]	6
S4.3 Chromosome 3q29 Microduplication syndrome	9
S4.4 WAGR syndrome [MIM:194072]	10
S4.5 Cri du chat syndrome [MIM:123450]	11
S4.6 Potocki Shaffer Syndrome [MIM:601224]	14
S4.7 Kleefstra syndrome [MIM:610253]	16
S4.8 Chromosome 1p36 Deletion Syndrome [MIM:607872]	18
S4.9 Angelman Syndrome [MIM:105830]	22
S4.10 Chromosome 1q21.1 deletion syndrome [MIM:274000]	24
S4.11 Potocki Lupski syndrome [MIM:610883]	25
S4.12 Renal Cysts and Diabetes (RCAD) syndrome [MIM:137920]	27
S4.13 Leri Weill Dyschondrosteosis [MIM:127300]	29
S4.14 Prader Willi Syndrome [MIM:176270]	30
S4.15 Split hand/foot malformation 1 [MIM:183600]	33
S4.16 Rubinstein Taybi Syndrome [MIM:180849]	34
S4.17 Chromosome 22q13.3 Deletion Syndrome [MIM:606232]	37
S4.18 Pelizaeus Merzbacher Disease [MIM:312080]	39
S4.19 Sotos syndrome [MIM:117550]	41
S4.20 Wolf Hirschhorn Syndrome [MIM:194190]	43
S4.21 15q26 Overgrowth Syndrome [DECIPHER:81]	46

S4.22 Chromosome 15q24 Deletion Syndrome [MIM:613406]	48
S4.23 Smith Magenis Syndrome [MIM:182290]	51
S4.24 Chromosome 17q11.2 deletion syndrome [MIM:613675]	54
S4.25 Xq28 (MECP2) Duplication [DECIPHER:45]	56
S4.26 Chromosome 17q21.31 Microdeletion Syndrome [MIM:610443]	58
S4.27 Adenomatous polyposis of the colon [MIM:175100]	60

S4 Annotations of CNV disorders

Comprehensive phenotypic annotations were generated for the 27 CNV disorders by manual curation. The following tables present the phenotypic annotations and the lists of genes contained within the CNV intervals. The references include primary literature, OMIM¹, Smith's *Recognizable Patterns of Human Malformation*², and entries from NCBI's GeneClinics. Note that entries from OMIM refer to the Webpage with the indicated MIM number.

S4.1 Miller Dieker Lissencephaly Syndrome [MIM:247200]

Term name	Term ID	Frequency	Ref.
Autosomal dominant inheritance	HP:0000006	100.00 %	1
Inguinal hernia	HP:0000023	100.00 %	1
Cryptorchidism	HP:0000028	100.00 %	1
Pelvic kidney	HP:0000125	100.00 %	1
Cleft palate	HP:0000175	100.00 %	1
Cataract	HP:0000518	100.00 %	1
Upstalting palpebral fissures	HP:0000582	100.00 %	1
Delayed eruption of teeth	HP:0000684	100.00 %	1
Motor delay	HP:0001270	100.00 %	1
Pachygyria	HP:0001302	100.00 %	1
Lissencephaly	HP:0001339	100.00 %	3
Failure to thrive	HP:0001508	100.00 %	1
Omphalocele	HP:0001539	100.00 %	1
Abnormality of metabolism/homeostasis	HP:0001939	100.00 %	1
Aspiration pneumonia	HP:0002100	100.00 %	1
Duodenal atresia	HP:0002247	100.00 %	1
Heterotopia	HP:0002282	100.00 %	1
Infantile seizures	HP:0002391	100.00 %	1
Nasal hypoplasia	HP:0003196	100.00 %	3
Progressive spastic paraplegia	HP:0007020	100.00 %	1
Decreased fetal movement	HP:0001558	100.00 %	1
Thick upper lip vermillion	HP:0000215	100.00 %	3
Intrauterine growth restriction	HP:0001511	100.00 %	1
Intellectual disability	HP:0001249	100.00 %	1
Micrognathia	HP:0000347	96.30 %	3
Broad nasal bridge	HP:0000431	96.00 %	3
Posteriorly rotated ears	HP:0000358	90.00 %	4
Frontal bossing	HP:0002007	90.00 %	4
Cavum septum pellucidum	HP:0002389	77.27 %	3
Hypoplasia of the corpus callosum	HP:0002079	73.91 %	3
Sacral dimple	HP:0000960	73.68 %	3
Microcephaly	HP:0000252	68.00 %	3
Deep palmar creases	HP:0006191	66.67 %	3
Midline brain calcifications	HP:0007045	54.17 %	3
Low-set ears	HP:0000369	51.85 %	3
Clinodactyly of the 5th finger	HP:0004209	41.67 %	3
Epicanthus	HP:0000286	36.36 %	3
Intrauterine growth restriction	HP:0001511	36.36 %	3
Polyhydramnios	HP:0001561	30.00 %	3
Cardiac malformation	HP:0002564	22.22 %	3
Joint contractures involving the joints of the hand	HP:0009473	22.22 %	3
Transverse palmar creases	HP:0000954	20.83 %	3

Table S1: Phenotypic annotations for the Miller Dieker syndrome [MIM:247200].

Gene Symbol	Entrez ID	Position
ABR	29	chr:17:906758-1090615
C17orf97	400566	chr:17:260117-264456
CRK	1398	chr:17:1325439-1359543
DPH1	1801	chr:17:1933430-1946724
FAM101B	359845	chr:17:289768-295731
FAM57A	79850	chr:17:635846-646074
GEMIN4	50628	chr:17:647660-655500
GLOD4	51031	chr:17:662548-685570
HIC1	3090	chr:17:1958392-1962980
METTL16	79066	chr:17:2319342-2415199
MNT	4335	chr:17:2287353-2304257
MYO1C	4641	chr:17:1367479-1396000
NXXN	64359	chr:17:702584-883009
PAFAH1B1	5048	chr:17:2496922-2588908
PITPNNA	5306	chr:17:1421282-1466109
PRPF8	10594	chr:17:1553922-1588175
RILP	83547	chr:17:1549443-1553391
RNMTL1	55178	chr:17:685512-695740
RPA1	6117	chr:17:1733272-1802847
RPH3AL	9501	chr:17:62179-202632
RTN4RL1	146760	chr:17:1837970-1928177
SCARF1	8578	chr:17:1537151-1549040
SERPINF1	5176	chr:17:1665258-1680867
SERPINF2	5345	chr:17:1646129-1658558
SLC43A2	124935	chr:17:1477695-1532129
SMG6	23293	chr:17:1963132-2207068
SMYD4	114826	chr:17:1682828-1733174
SNORD91A	692207	chr:17:2233572-2233663
SNORD91B	692208	chr:17:2232418-2232503
SRR	63826	chr:17:2207247-2228552
TIMM22	29928	chr:17:900356-905406
TLCD2	727910	chr:17:1606083-1613661
TSR1	55720	chr:17:2225971-2240677
TUSC5	286753	chr:17:1182956-1204280
VPS53	55275	chr:17:411907-618095
WDR81	124997	chr:17:1619816-1641892
YWHAE	7531	chr:17:1247833-1303555

Table **S2:** Genes located within the Miller Dieker interval [MIM:247200].

S4.2 Williams Beuren Syndrome [MIM:194050]

Term name	Term ID	Frequency	Reference
Autosomal dominant inheritance	HP:0000006	100.00 %	5
Anxiety	HP:0000739	80.00 %	5
Recurrent urinary tract infections	HP:0000010	75.00 %	5
Hypodontia	HP:0000668	75.00 %	5
Intellectual disability	HP:0001249	75.00 %	5
Short stature	HP:0004322	75.00 %	5
Mitral valve prolapse	HP:0001634	75.00 %	5
Pulmonic stenosis	HP:0001642	75.00 %	6
Mitral regurgitation	HP:0001653	75.00 %	5
Constipation	HP:0002019	75.00 %	5
Supravalvular aortic stenosis	HP:0004381	75.00 %	6
peripheral pulmonary artery stenosis	HP:0004969	75.00 %	5
Coronary artery stenosis	HP:0005145	75.00 %	5
Hyperacusis	HP:0010780	75.00 %	5
Recurrent otitis media	HP:0000403	75.00 %	5
Diabetes mellitus	HP:0000819	75.00 %	5
Osteopenia	HP:0000938	75.00 %	5
Osteoporosis	HP:0000939	75.00 %	5
Gastroesophageal reflux	HP:0002020	75.00 %	5
Rectal prolapse	HP:0002035	75.00 %	5
Kidney malformation	HP:0000792	75.00 %	5
Contractures	HP:0001371	75.00 %	5
Joint laxity	HP:0001388	75.00 %	5
Muscular hypotonia	HP:0001252	75.00 %	5
Hyperreflexia	HP:0001347	75.00 %	5
Strabismus	HP:0000486	75.00 %	5
Premature graying of hair	HP:0002216	75.00 %	5
Failure to thrive in infancy	HP:0001531	70.00 %	6
Full lips	HP:0000170	50.00 %	2
Epicanthus	HP:0000286	50.00 %	2
Flattened nasal bridge	HP:0000425	50.00 %	2
Feeding problems in infancy	HP:0008872	50.00 %	2
Hoarse voice	HP:0001609	50.00 %	2
Nail hypoplasia	HP:0001792	50.00 %	2
Hallux valgus	HP:0001822	50.00 %	2
Phonophobia	HP:0002183	50.00 %	5
Urethral stenosis	HP:0008661	50.00 %	2
Early onset of puberty	HP:0100000	50.00 %	6
Blepharophimosis	HP:0000581	50.00 %	2
Medial flaring of the eyebrow	HP:0010747	50.00 %	2
Periorbital fullness	HP:0000629	50.00 %	2
Blue irides	HP:0000635	50.00 %	2
Nares, anteverted	HP:0000463	50.00 %	2
Long philtrum	HP:0000343	50.00 %	2
Open mouth	HP:0000194	50.00 %	2
Soft skin	HP:0000977	50.00 %	2
Obesity	HP:0001513	50.00 %	2
Cutis laxa	HP:0000973	50.00 %	2
Bitemporal narrowing	HP:0000314	50.00 %	6

Term name	Term ID	Frequency	Reference
Nasal hypoplasia	HP:0003196	50.00 %	6
Broad nasal tip	HP:0000455	50.00 %	6
Long philtrum	HP:0000343	50.00 %	6
Prominent ear lobes	HP:0004456	50.00 %	6
Full cheeks	HP:0000293	50.00 %	2
Sloping shoulders	HP:0001556	50.00 %	2
Impaired visuospatial constructive cognition	HP:0010794	50.00 %	5
Bladder diverticula	HP:0000015	33.00 %	5
Microdontia	HP:0000691	33.00 %	5
Kyphoscoliosis	HP:0002751	33.00 %	5
Attention deficit hyperactivity disorder	HP:0007018	33.00 %	5
Sensorineural hearing impairment	HP:0000407	33.00 %	5
Dental malocclusion	HP:0000689	33.00 %	5
Gait imbalance	HP:0002141	33.00 %	5
Incoordination	HP:0002311	33.00 %	5
Colon diverticula	HP:0002253	30.00 %	5
Hypercalciuria	HP:0002150	30.00 %	6
Myxomatous mitral valve degeneration	HP:0004764	20.00 %	5
Hypercalcemia	HP:0003072	15.00 %	6
Cerebellar hypoplasia	HP:0001321	15.00 %	5
Nephrocalcinosis	HP:0000121	10.00 %	5
Hypothyroidism	HP:0000821	10.00 %	6
Arnold-Chiari type I malformation	HP:0007099	10.00 %	5
Inguinal hernia	HP:0000023	7.50 %	2
Pectus excavatum	HP:0000767	7.50 %	2
Ventricular septal defect	HP:0001629	7.50 %	5
Atrial septal defect	HP:0001631	7.50 %	5
Celiac disease	HP:0002608	7.50 %	5
Sleep disturbances	HP:0002360	7.50 %	5
Vocal cord paralysis	HP:0001605	7.50 %	2
Hypotelorism	HP:0000601	7.50 %	2
Amblyopia	HP:0000646	7.50 %	2
Abnormality of refraction	HP:0000539	7.50 %	2
Retinal arteriolar tortuosity	HP:0001136	7.50 %	2
Malar hypoplasia	HP:0000272	7.50 %	2
Clinodactyly of the 5th finger	HP:0004209	7.50 %	2
Radioulnar synostosis	HP:0002974	7.50 %	2
Micropenis	HP:0000054	7.50 %	2
Umbilical hernia	HP:0001537	7.50 %	2
Portal hypertension	HP:0001409	7.50 %	2
Renal artery stenosis	HP:0001920	5.00 %	5
Stroke	HP:0001297	1.00 %	5
Sudden death	HP:0001699	1.00 %	5

Table S3: Phenotypic annotations for Williams Beuren Syndrome [MIM:194050].

Gene Symbol	Entrez ID	Position
TRIM74	378108	chr:7:72430015-72439996
NSUN5	55695	chr:7:72716513-72722863
TRIM50	135892	chr:7:72726534-72742084
FKBP6	8468	chr:7:72742154-72772640
FZD9	8326	chr:7:72848108-72850449
BAZ1B	9031	chr:7:72854727-72936614
BCL7B	9275	chr:7:72950685-72972023
TBL2	26608	chr:7:72983273-72993012
MLXIPL	51085	chr:7:73007523-73038869
VPS37D	155382	chr:7:73082173-73086439
WBSCR22	114049	chr:7:73097897-73112541
STX1A	6804	chr:7:73113534-73134016
ABHD11	83451	chr:7:73150423-73153189
CLDN3	1365	chr:7:73183326-73184599
CLDN4	1364	chr:7:73245192-73247014
WBSCR27	155368	chr:7:73248919-73256854
WBSCR28	135886	chr:7:73275488-73280222
ELN	2006	chr:7:73442426-73484236
LIMK1	3984	chr:7:73498155-73536853
EIF4H	7458	chr:7:73588705-73611428
LAT2	7462	chr:7:73624086-73644163
RFC2	5982	chr:7:73645831-73668737
CLIP2	7461	chr:7:73703804-73820272
GTF2IRD1	9569	chr:7:73868119-74016916
GTF2I	2969	chr:7:74072029-74175021
NCF1	653361	chr:7:74188308-74203658
GTF2IRD2	84163	chr:7:74210483-74267840
WBSCR16	81554	chr:7:74456282-74489698
GTF2IRD2B	389524	chr:7:74508346-74565622
GTF2IRD2P	401375	chr:7:72656902-72694143
NCF1C	654817	chr:7:74572445-74587848
NSUN5B	155400	chr:7:72418832-72425329
NSUN5C	260294	chr:7:72418832-72425329
PMS2L5	5383	chr:7:74306894-74366314

Table S4: Genes in the Williams syndrome interval [MIM:194050].

S4.3 Chromosome 3q29 Microduplication syndrome

Term name	Term ID	Frequency	Ref.
Autosomal dominant inheritance	HP:0000006	100.00 %	¹
Microcephaly	HP:0000252	80.00 %	⁷
Round face	HP:0000311	75.00 %	⁷
Intellectual disability	HP:0001249	75.00 %	⁷
Bulbous nose	HP:0000414	75.00 %	⁷
Obesity	HP:0001513	60.00 %	⁷
Nasal hypoplasia	HP:0003196	50.00 %	⁷
Multiple palmar creases	HP:0006114	50.00 %	⁷
Pes planus	HP:0001763	50.00 %	⁷
Long face	HP:0000276	25.00 %	⁷
Downward slanting palpebral fissures	HP:0000494	25.00 %	⁷
Blepharophimosis	HP:0000581	25.00 %	⁷
Broad nasal bridge	HP:0000431	25.00 %	⁷
Low posterior hairline	HP:0002162	25.00 %	⁷
Macrocephaly	HP:0000256	20.00 %	⁷

Table S5: Phenotypic annotations for Chromosome 3q29 Microduplication syndrome [MIM:611936].

Gene Symbol	Entrez ID	Position
BDH1	622	chr:3:197236653-197300193
KIAA0226	9711	chr:3:197398258-197476567
FYTTD1	84248	chr:3:197476423-197511316
CEP19	84984	chr:3:196433147-196439122
C3orf43	255798	chr:3:196233749-196242236
DLG1	1739	chr:3:196769430-197025446
FBXO45	200933	chr:3:196295724-196315929
LRRC33	375387	chr:3:196366655-196388874
MFI2	4241	chr:3:196728610-196756686
NCBP2	22916	chr:3:196662272-196669463
PAK2	5062	chr:3:196466727-196559517
PCYT1A	5130	chr:3:195965252-196014583
PIGX	54965	chr:3:196439244-196462877
PIGZ	80235	chr:3:196673213-196695703
RNF168	165918	chr:3:196195653-196230638
SENP5	205564	chr:3:196594726-196661584
TCTEX1D2	255758	chr:3:196018089-196045158
TFRC	7037	chr:3:195776154-195809031
TM4SF19	116211	chr:3:196050418-196065257
UBXN7	26043	chr:3:196080360-196159344
WDR53	348793	chr:3:196281058-196295412
ZDHHC19	131540	chr:3:195924322-195938299

Table S6: Genes in the chromosome 3q29 Microduplication syndrome interval.

S4.4 Wilms tumor, aniridia, genitourinary anomalies and mental retardation (WAGR) syndrome [MIM:194072]

Term name	Term ID	Frequency	Reference
Autosomal dominant inheritance	HP:0000006	100.00 %	MIM:194072
Aniridia	HP:0000526	90.00 %	MIM:194072
Cryptorchidism	HP:0000028	50.00 %	MIM:194072
Hypospadias	HP:0000047	50.00 %	MIM:194072
Intellectual disability	HP:0001249	50.00 %	MIM:194072
Nephroblastoma (Wilms tumor)	HP:0002667	50.00 %	MIM:194072
Nephropathy	HP:0000112	40.00 %	⁸
Abnormality of the vagina	HP:0000142	33.00 %	⁸
Streak ovary	HP:0010464	33.00 %	⁸
Renal failure	HP:0000083	21.74 %	⁹
Abnormality of the uterus	HP:0000130	7.50 %	⁸
Obesity	HP:0001513	7.50 %	⁸
Gonadoblastoma	HP:0000150	7.50 %	⁸
Obesity	HP:0001513	7.50 %	MIM:194072

Gene Symbol	Entrez ID	Position
ELP4	26610	chr:11:31531296-31805328
PAX6	5080	chr:11:31806339-31839508
RCN1	5954	chr:11:32112476-32127271
WT1	7490	chr:11:32409324-32457086
WT1-AS	51352	chr:11:32457284-32461635

Table S8: WAGR syndrome [MIM:194072].

S4.5 Cri du chat syndrome [MIM:123450]

Term name	Term ID	Frequency	Reference
Sporadic	HP:0003745	100.00 %	2
Microcephaly	HP:0000252	100.00 %	2
Growth retardation, prenatal and postnatal	HP:0008893	100.00 %	2
Intellectual disability	HP:0001249	100.00 %	2
Microretrognathia	HP:0000308	97.00 %	10
Cat cry	HP:0200046	96.00 %	11
Hypertelorism	HP:0000316	94.00 %	2
Speech and language difficulties	HP:0002399	90.00 %	12
Short philtrum	HP:0000322	88.00 %	11
Broad nasal bridge	HP:0000431	87.00 %	11
Epicanthus	HP:0000286	85.00 %	2
Hypoplastic/short metacarpal bones	HP:0010049	83.00 %	11
Stereotyped, repetitive behaviour	HP:0000733	82.00 %	13
Difficulty walking	HP:0002355	81.48 %	14
Downward slanting palpebral fissures	HP:0000494	81.00 %	2
Transverse palmar creases	HP:0000954	81.00 %	2
Downturned corners of mouth	HP:0002714	81.00 %	11
Neonatal hypotonia	HP:0001319	78.00 %	2
Round face	HP:0000311	75.00 %	2,11
Anterior open-bite malocclusion	HP:0009102	75.00 %	11
Hypoplasia of the metatarsal bones	HP:0010743	75.00 %	11
Short attention span	HP:0000736	75.00 %	10
Anxiety	HP:0000739	75.00 %	10
Low birth weight	HP:0001518	72.00 %	2
Long, narrow facies	HP:0000318	71.00 %	11
Self-mutilation	HP:0000742	70.37 %	14
Conspicuously happy disposition	HP:0100024	66.67 %	14
Strabismus	HP:0000486	61.00 %	2
Low-set ears	HP:0000369	58.00 %	2
Poorly formed pinnae	HP:0008562	58.00 %	2
Echolalia	HP:0010529	51.85 %	14
Facial asymmetry	HP:0000324	50.00 %	2
High palate	HP:0000218	50.00 %	10
Hyperactivity	HP:0000752	50.00 %	10,11
Overfriendliness	HP:0100025	48.15 %	14
Thick lower lip vermillion	HP:0000179	45.00 %	11
High axial triradius	HP:0001042	40.00 %	2
Autism	HP:0000717	39.13 %	15
Facial grimacing	HP:0000273	33.33 %	14
Oppositional defiant disorder	HP:0010865	33.33 %	14
Feeding problems in infancy	HP:0008872	33.00 %	11
Gastroesophageal reflux	HP:0002020	33.00 %	11
Diastasis recti	HP:0001540	33.00 %	11
Functional respiratory abnormality	HP:0002795	33.00 %	10
Hyperacusis	HP:0010780	33.00 %	10
Hypertonia	HP:0001276	33.00 %	10
Prominent supraorbital ridges	HP:0000336	31.00 %	11
Cardiac malformation	HP:0002564	30.00 %	2

Term name	Term ID	Frequency	Reference
Premature graying of hair	HP:0002216	30.00 %	11
Recurrent infections in infancy and early childhood	HP:0005437	7.50 %	11
Hypospadias	HP:0000047	7.50 %	11
Cryptorchidism	HP:0000028	7.50 %	11
Preauricular skin tag	HP:0000384	7.50 %	11
Syndactyly	HP:0001159	7.50 %	11
Myopia	HP:0000545	7.50 %	2
Optic atrophy	HP:0000648	7.50 %	2
Bifid uvula	HP:0000193	7.50 %	2
Short neck	HP:0000470	7.50 %	2
Scoliosis	HP:0002650	7.50 %	2
Pes planus	HP:0001763	7.50 %	2
Inguinal hernia	HP:0000023	7.50 %	2
Metatarsus varus	HP:0001840	7.50 %	2
Stenotic external auditory canal	HP:0000402	7.50 %	10
Hyperacusis	HP:0010780	7.50 %	10
Deafness	HP:0000404	7.50 %	10
Cataract	HP:0000518	7.50 %	10
Abnormality of the kidney	HP:0000077	5.00 %	10,11
Cleft lip/palate	HP:0000202	5.00 %	10

Gene Symbol	Entrez ID	Position
IRX1	79192	chr:5:3596167-3601516
ADAMTS16	170690	chr:5:5140442-5320411
MED10	84246	chr:5:6372038-6378638
NSUN2	54888	chr:5:6599351-6633472
SRD5A1	6715	chr:5:6633499-6669674
PAPD7	11044	chr:5:6714717-6757160
ADCY2	108	chr:5:7396342-7830193
MTRR	4552	chr:5:7869216-7901236
FASTKD3	79072	chr:5:7859271-7869149
SEMA5A	9037	chr:5:9035137-9546232
TAS2R1	50834	chr:5:9629108-9630462
CCT5	22948	chr:5:10250281-10266500
CMBL	134147	chr:5:10277706-10308167
MARCH6	10299	chr:5:10353827-10435490
ROPN1L	83853	chr:5:10442008-10465137
DAP	1611	chr:5:10679341-10761386
CTNND2	1501	chr:5:10971951-11904109
PLEKHG4B	153478	chr:5:140372-190086
CCDC127	133957	chr:5:204874-218296
SDHA	6389	chr:5:218355-256814
PDCD6	10016	chr:5:271735-315088
EXOC3	11336	chr:5:443333-467410
SLC9A3	6550	chr:5:473333-524548
CEP72	55722	chr:5:612404-653667
TPPP	11076	chr:5:659976-693509

Gene Symbol	Entrez ID	Position
ZDHHC11	79844	chr:5:795719-851100
BRD9	65980	chr:5:863849-892938
TRIP13	9319	chr:5:892968-918163
NKD2	85409	chr:5:1009167-1038924
SLC12A7	10723	chr:5:1050488-1112171
SLC6A19	340024	chr:5:1201709-1225231
SLC6A18	348932	chr:5:1225469-1246303
TERT	7015	chr:5:1253281-1295161
CLPTM1L	81037	chr:5:1317998-1345001
SLC6A3	6531	chr:5:1392904-1445542
LPCAT1	79888	chr:5:1461541-1524075
MRPL36	64979	chr:5:1798498-1799955
NDUFS6	4726	chr:5:1801495-1816164
IRX4	50805	chr:5:1877540-1882879
IRX2	153572	chr:5:2746278-2751768
C5orf38	153571	chr:5:2752261-2755510
UBE2QL1	134111	chr:5:6448735-6492705

Table S10: Cri du chat syndrome [MIM:123450].

S4.6 Potocki Shaffer Syndrome [MIM:601224]

Also known as Chromosome 11p11.2 deletion syndrome

Term name	Term ID	Frequency	Ref.
Turricephaly	HP:0000262	100.00 %	¹
Contiguous gene syndrome	HP:0001466	100.00 %	¹
Multiple exostoses	HP:0002762	100.00 %	¹⁶
Broad forehead	HP:0000337	100.00 %	¹⁶
High forehead	HP:0000348	100.00 %	¹⁶
Downward slanting palpebral fissures	HP:0000494	100.00 %	¹⁶
Broad nasal bridge	HP:0000431	100.00 %	¹⁶
Nasal hypoplasia	HP:0003196	100.00 %	¹⁶
Hypoplastic nasal alae	HP:0000430	100.00 %	¹⁶
Downturned corners of mouth	HP:0002714	88.89 %	¹⁷
Transverse palmar creases	HP:0000954	83.33 %	¹⁶
Micropenis	HP:0000054	83.33 %	¹⁶
Parietal foramina	HP:0002697	81.82 %	¹⁶
Intellectual disability	HP:0001249	70.00 %	¹⁶
Brachycephaly	HP:0000248	66.67 %	¹⁶
Sparse lateral eyebrows	HP:0005338	66.67 %	¹⁷
Short philtrum	HP:0000322	66.67 %	¹⁷
Brachydactyly	HP:0001156	62.50 %	¹⁷
Muscular hypotonia	HP:0001252	55.56 %	¹⁶
Wormian bones	HP:0002645	50.00 %	¹⁶
Telecanthus	HP:0000506	44.44 %	¹⁶
Epicanthus	HP:0000286	44.44 %	¹⁶
Craniofacial dysostosis	HP:0004439	33.00 %	¹⁶
Seizures	HP:0001250	18.18 %	¹⁶
Cutaneous syndactyly between fingers	HP:0005650	5.00 %	¹
2 and 5			

Table S11: Phenotypic annotations for Potocki Shaffer Syndrome [MIM:601224].

Gene Symbol	Entrez ID	Position
ACCS	84680	chr:11:44087728-44105568
EXT2	2132	chr:11:44117098-44266979
ALX4	60529	chr:11:44282277-44331715
CD82	3732	chr:11:44587140-44641338
TSPAN18	90139	chr:11:44785975-44953977
TP53I11	9537	chr:11:44953898-44972607
PRDM11	56981	chr:11:45115563-45246902
SYT13	57586	chr:11:45261852-45307883
CHST1	8534	chr:11:45670426-45687171
SLC35C1	55343	chr:11:45825622-45834566
CRY2	1408	chr:11:45868668-45904798
MAPK8IP1	9479	chr:11:45907201-45928015

Gene Symbol	Entrez ID	Position
PEX16	9409	chr:11:45931219-45939673
GYLTL1B	120071	chr:11:45943195-45950646
PHF21A	51317	chr:11:45950869-46142984

Table **S12:** Genes in the Potocki Shaffer Syndrome interval [MIM:601224].

S4.7 Kleefstra syndrome [MIM:610253]

Also known as 9q Subtelomeric Deletion Syndrome, 9q34.3 Microdeletion Syndrome, 9qSTDS, Chromosome 9q34.3 Deletion Syndrome.

Term name	Term ID	Frequency	Ref.
Autosomal dominant inheritance	HP:0000006	100.00 %	¹⁸
Isolated cases	HP:0001420	100.00 %	¹⁸
Muscular hypotonia	HP:0001252	90.00 %	¹⁸
Intellectual disability	HP:0001249	90.00 %	¹⁸
Mandibular prognathia	HP:0000303	50.00 %	¹⁸
Abnormality of the pinna	HP:0000377	50.00 %	¹⁸
Upstalting palpebral fissures	HP:0000582	50.00 %	¹⁸
Impaired language development	HP:0000750	50.00 %	¹⁸
Coarse facial features	HP:0000280	50.00 %	¹⁸
Conotruncal defect	HP:0001710	50.00 %	¹⁸
Brachycephaly	HP:0000248	50.00 %	¹⁸ s
Malar hypoplasia	HP:0000272	50.00 %	¹⁸
Hypertelorism	HP:0000316	50.00 %	¹⁸
Synophrys	HP:0000664	50.00 %	¹⁸
Nares, antverted	HP:0000463	50.00 %	¹⁸
Everted lower lip vermillion	HP:0000232	50.00 %	¹⁸
U-Shaped upper lip vermillion	HP:0010806	50.00 %	¹⁸
Macroglossia	HP:0000158	50.00 %	¹⁸
Protruding tongue	HP:0010808	50.00 %	¹⁸
Microcephaly	HP:0000252	36.36 %	¹⁹
Aggressive behavior	HP:0000718	33.00 %	¹
Obesity	HP:0001513	33.00 %	¹⁹
Sleep disturbances	HP:0002360	33.00 %	¹⁸
Cryptorchidism	HP:0000028	33.00 %	¹⁸
Hypospadias	HP:0000047	33.00 %	¹⁸
Micropenis	HP:0000054	33.00 %	¹⁸
Stereotyped, repetitive behaviour	HP:0000733	33.00 %	¹⁸
Recurrent respiratory infections	HP:0002205	33.00 %	¹⁸
Autism	HP:0000717	33.00 %	¹⁸
Seizures	HP:0001250	30.00 %	¹⁸
Kidney malformation	HP:0000792	15.00 %	¹⁸
Hearing impairment	HP:0000365	13.64 %	¹⁹
Natal tooth	HP:0000695	7.50 %	¹⁸
Persistence of primary teeth	HP:0006335	7.50 %	¹⁸
Talipes equinovarus	HP:0001762	7.50 %	¹⁸
Gastroesophageal reflux	HP:0002020	5.00 %	¹⁸
Tracheobronchomalacia	HP:0002786	1.00 %	¹⁸
Apathy	HP:0000741	1.00 %	¹⁸

Table S13: Phenotypic annotations for the Kleefstra syndrome [MIM:610253].

Gene Symbol	Entrez ID	Position
ARRDC1	92714	chr:9:140500095-140509811
C9orf37	85026	chr:9:140509783-140513307
CACNA1B	774	chr:9:140772240-141019075
EHMT1	79813	chr:9:140605416-140730578
MRPL41	64975	chr:9:140446308-140447006
PNPLA7	375775	chr:9:140354404-140444985
WDR85	92715	chr:9:140449360-140473386
ZMYND19	116225	chr:9:140476530-140484936

Table **S14**: Genes in the Kleefstra syndrome [MIM:610253] interval.

S4.8 Chromosome 1p36 Deletion Syndrome [MIM:607872]

Term name	Term ID	Frequency	Ref.
Isolated cases	HP:0001420	100.00 %	²⁰
Impaired language development	HP:0000750	99.00 %	^{21,22}
Intellectual disability	HP:0001249	97.50 %	^{21,22}
Muscular hypotonia	HP:0001252	95.00 %	^{20,22}
Malformation of the central nervous system	HP:0007319	88.00 %	²²
Camptodactyly (hands)	HP:0100490	80.00 %	²²
Short feet	HP:0001766	80.00 %	²²
Wide anterior fontanel	HP:0000260	77.00 %	²⁰
Delayed closure of the anterior fontanelle	HP:0001476	77.00 %	²⁰
Pointed chin	HP:0000307	75.00 %	²
Flattened nasal bridge	HP:0000425	75.00 %	^{21,23}
Deeply set eye	HP:0000490	75.00 %	^{21,23}
Feeding problems in infancy	HP:0008872	70.00 %	^{21,23}
Flat nose	HP:0000457	67.00 %	²³
Strabismus	HP:0000486	67.00 %	²¹
Constipation	HP:0002019	65.00 %	²¹
Microcephaly	HP:0000252	61.00 %	²¹
Dysphagia	HP:0002015	60.50 %	^{21,23}
Sensorineural hearing impairment	HP:0000407	58.00 %	²¹
Thickened helices	HP:0000391	58.00 %	²¹
Epilepsy	HP:0001275	58.00 %	²⁴
Gastroesophageal reflux	HP:0002020	56.00 %	²¹
Self-mutilation	HP:0000742	55.00 %	^{2,20}
Brachycephaly	HP:0000248	54.00 %	²¹
Impaired vision	HP:0000505	54.00 %	^{21,22}
Impaired social interactions	HP:0000735	52.00 %	²²
Malar hypoplasia	HP:0000272	51.00 %	²¹
Hypermetropia	HP:0000540	50.00 %	^{21,23}
Clinodactyly of the 5th finger	HP:0004209	50.00 %	^{23,21}
Hypoplastic/small 5th finger	HP:0009237	50.00 %	²
Growth delay	HP:0001510	50.00 %	²
Obesity	HP:0001513	50.00 %	²
Frontal bossing	HP:0002007	50.00 %	²
Epicanthus	HP:0000286	50.00 %	²²
Hyperphagia	HP:0000724	50.00 %	²⁵
Low-set ears	HP:0000369	49.00 %	²¹
Hypoplasia of the external ear	HP:0008551	48.00 %	²¹
Abnormality of the hairline	HP:0009553	48.00 %	²¹
Conductive hearing impairment	HP:0000405	46.00 %	²¹
Myopia	HP:0000545	40.00 %	^{21,23}
Asymmetry of the ears	HP:0010722	40.00 %	²¹
Hypertelorism	HP:0000316	38.00 %	²¹
Dilatation of lateral cerebral ventricles	HP:0006796	31.50 %	^{21,22}
Ventricular septal defect	HP:0001629	29.00 %	²¹
Leukoencephalopathy	HP:0002352	28.00 %	²²
Dilated cardiomyopathy	HP:0001644	27.00 %	²¹⁻²³
Blepharophimosis	HP:0000581	27.00 %	²¹

Term name	Term ID	Frequency	Ref.
Upstenting palpebral fissures	HP:0000582	27.00 %	21
Downward slanting palpebral fissures	HP:0000494	26.00 %	21
Hypsarrhythmia	HP:0002521	25.00 %	20
Patent foramen ovale	HP:0001655	24.00 %	21
Posteriorly rotated ears	HP:0000358	23.00 %	23
Patent ductus arteriosus	HP:0001643	22.00 %	21,23
Abnormality of the kidney	HP:0000077	22.00 %	22
Oppositional defiant disorder	HP:0010865	22.00 %	22
Delayed skeletal maturation	HP:0002750	22.00 %	22
Synophrys	HP:0000664	21.00 %	21
Cerebral cortical atrophy	HP:0002120	20.00 %	22
Polymicrogyria	HP:0002126	20.00 %	21
Congenital hypothyroidism	HP:0000851	17.50 %	22,23
Scoliosis	HP:0002650	16.00 %	22
Nystagmus	HP:0000639	13.00 %	2,23
Thin corpus callosum	HP:0002319	12.00 %	22
Cryptorchidism	HP:0000028	10.00 %	22
Aortic root dilatation	HP:0002616	10.00 %	23
Atrial septal defect	HP:0001631	8.00 %	21,23
Delayed myelination	HP:0002188	8.00 %	22
Bifid uvula	HP:0000193	7.50 %	2
Cleft lip	HP:0000204	7.50 %	2
Hydrocephalus	HP:0000238	7.50 %	2
Cranial nerve VI palsy	HP:0006897	7.50 %	2
Optic atrophy, congenital	HP:0007855	7.50 %	2
Long philtrum	HP:0000343	7.50 %	2
Metatarsus varus	HP:0001840	7.50 %	2
11 pairs of ribs	HP:0000878	7.50 %	22
Bifid ribs	HP:0000892	7.50 %	22
Rib fusion	HP:0000902	7.50 %	22
Cataract	HP:0000518	6.00 %	22
Submucous cleft lip/palate	HP:0000208	5.00 %	23
Bicuspid aortic valve	HP:0001647	5.00 %	21,23
Abnormal lung lobation	HP:0002101	5.00 %	2
Hypospadias	HP:0000047	5.00 %	22
Ebstein's anomaly of the tricuspid valve	HP:0010316	3.00 %	21,23
Optic nerve coloboma	HP:0000588	3.00 %	22
Hip dysplasia	HP:0001385	3.00 %	2
Abnormality of the anus	HP:0004378	3.00 %	22

Table S15: Phenotypic annotations for the Chromosome 1p36 Deletion Syndrome [MIM:607872].

Gene Symbol	Entrez ID	Position
ACTRT2	140625	chr:1:2938045-2939466
AGRN	375790	chr:1:955502-991491
AJAP1	55966	chr:1:4715104-4843850
ARHGEF16	27237	chr:1:3371146-3397676

Gene Symbol	Entrez ID	Position
ATAD3A	55210	chr:1:1447522-1470066
ATAD3B	83858	chr:1:1407163-1431581
ATAD3C	219293	chr:1:1385068-1405537
AURKAIP1	54998	chr:1:1309109-1310817
B3GALT6	126792	chr:1:1167628-1170420
C1orf159	54991	chr:1:1017197-1051735
C1orf170	84808	chr:1:910578-917472
C1orf174	339448	chr:1:3805696-3816856
KIAA1751	85452	chr:1:1884751-1935275
C1orf70	339453	chr:1:1470157-1475739
C1orf86	199990	chr:1:2115898-2139171
C1orf93	127281	chr:1:2518248-2522901
CALML6	163688	chr:1:1846265-1848732
CCDC27	148870	chr:1:3668964-3688208
CCNL2	81669	chr:1:1321090-1334717
CDK11A	728642	chr:1:1634168-1655790
ACAP3	116983	chr:1:1227763-1243268
CPSF3L	54973	chr:1:1246964-1260045
DFFB	1677	chr:1:3773844-3801992
DVL1	1855	chr:1:1270657-1284491
FAM132A	388581	chr:1:1177832-1182101
GABRD	2563	chr:1:1950767-1962191
GNB1	2782	chr:1:1716728-1822494
HES4	57801	chr:1:934341-935551
HES5	388585	chr:1:2460183-2461683
ISG15	9636	chr:1:948846-949919
CEP104	9731	chr:1:3728644-3773796
KIAA1751	85452	chr:1:1884751-1935275
KLHL17	339451	chr:1:895966-901094
LRRC47	57470	chr:1:3696783-3713067
MEGF6	1953	chr:1:3404505-3528058
MIB2	142678	chr:1:1550794-1565989
MMEL1	79258	chr:1:2522080-2564480
MMP23B	8510	chr:1:1567559-1570029
MORN1	79906	chr:1:2252695-2322992
MRPL20	55052	chr:1:1337275-1342692
MXRA8	54587	chr:1:1288070-1293914
NADK	65220	chr:1:1682670-1709908
NOC2L	26155	chr:1:879582-894678
OR4F29	729759	chr:1:367658-368596
OR4F5	79501	chr:1:69090-70007
PANK4	55229	chr:1:2439974-2458034
PEX10	5192	chr:1:2336240-2344009
PLCH2	9651	chr:1:2407753-2436963
PLEKHN1	84069	chr:1:901876-910487
PRDM16	63976	chr:1:2985741-3355184
PRKCZ	5590	chr:1:1981908-2116833
PUSL1	126789	chr:1:1243993-1247056
RER1	11079	chr:1:2323213-2336882
SAMD11	148398	chr:1:861120-879960
SCNN1D	6339	chr:1:1217488-1227408
SDF4	51150	chr:1:1152287-1167446

Gene Symbol	Entrez ID	Position
SKI	6497	chr:1:2160133-2241651
SLC35E2	9906	chr:1:1663680-1677430
SSU72	29101	chr:1:1477052-1510261
TAS1R3	83756	chr:1:1266725-1269843
TMEM52	339456	chr:1:1849028-1850739
TNFRSF14	8764	chr:1:2487804-2495267
TNFRSF18	8784	chr:1:1138887-1142088
TNFRSF4	7293	chr:1:1146705-1149511
TP73	7161	chr:1:3569128-3650466
TPRG1L	127262	chr:1:3541555-3546694
TTLL10	254173	chr:1:1109285-1133312
UBE2J2	118424	chr:1:1189291-1209233
VWA1	64856	chr:1:1370902-1378261
WRAP73	49856	chr:1:3547330-3566670

Table **S16**: Genes located in the Chromosome 1p36 Deletion Syndrome [MIM:607872] interval.

S4.9 Angelman Syndrome [MIM:105830]

Term name	Term ID	Frequency	Reference
Absent speech development	HP:0001344	100.00 %	2
Motor delay	HP:0001270	100.00 %	2
Intellectual disability, progressive	HP:0006887	100.00 %	2
Progressive gait ataxia	HP:0007240	100.00 %	2
EEG abnormalities	HP:0002353	92.00 %	2
Broad-based gait	HP:0002136	90.00 %	
Clumsiness	HP:0002312	90.00 %	
Blue irides	HP:0000635	88.00 %	2
Seizures	HP:0001250	86.00 %	2
Paroxysmal bursts of laughter	HP:0000749	75.00 %	1
Isolated cases	HP:0001420	75.00 %	
Flat occiput	HP:0005469	75.00 %	1
Microcephaly, postnatal	HP:0005484	75.00 %	1
Hyperactivity	HP:0000752	75.00 %	1
Blond hair	HP:0002214	65.00 %	2
Strabismus	HP:0000486	42.00 %	2
Hypopigmentation of the skin	HP:0001010	39.00 %	2
Mild cortical atrophy on ct or mri	HP:0006823	33.00 %	2
Macroglossia	HP:0000158	25.00 %	2
Wide mouth	HP:0000154	25.00 %	2
Mandibular prognathia	HP:0000303	25.00 %	2
Hypoplasia of the maxilla	HP:0000327	25.00 %	2
Deeply set eye	HP:0000490	25.00 %	2
Widely spaced teeth	HP:0000687	25.00 %	2
Muscular hypotonia	HP:0001252	25.00 %	2
Microbrachycephaly	HP:0002258	25.00 %	2
Nystagmus	HP:0000639	7.50 %	2
Hyperreflexia	HP:0001347	7.50 %	2
Obesity	HP:0001513	7.50 %	1
Constipation	HP:0002019	7.50 %	1
Feeding difficulties	HP:0002022	7.50 %	1
Scoliosis	HP:0002650	7.50 %	2
Sleep-wake cycle disturbance	HP:0006979	7.50 %	1
Autosomal dominant inheritance	HP:0000006	5.00 %	1

Table S17: Phenotypic annotations for Angelman syndrome [MIM:105830].

Gene Symbol	Entrez ID	Position
ATP10A	57194	chr:15:25923858-26108348
C15orf2	23742	chr:15:24920540-24928592
CYFIP1	23191	chr:15:22892683-23003602
GABRA5	2558	chr:15:27111865-27194356
GABRB3	2562	chr:15:26788692-27018934
GABRG3	2567	chr:15:27216428-27778372

Gene Symbol	Entrez ID	Position
HERC2	8924	chr:15:28356185-28567294
MKRN3	7681	chr:15:23810453-23813166
NDN	4692	chr:15:23930553-23932449
NIPA1	123606	chr:15:23043278-23086842
NIPA2	81614	chr:15:23004683-23034426
OCA2	4948	chr:15:28000020-28344457
SNORD107	91380	chr:15:25227140-25227214
SNORD109B	338429	chr:15:25523489-25523555
SNORD115-1	338433	chr:15:25415869-25415950
SNORD116-1	100033413	chr:15:25296622-25296718
SNORD116-10	100033422	chr:15:25319259-25319362
SNORD116-11	100033423	chr:15:25321074-25321167
SNORD116-12	100033424	chr:15:25322196-25322289
SNORD116-13	100033425	chr:15:25324203-25324296
SNORD116-14	100033426	chr:15:25325287-25325380
SNORD116-15	100033427	chr:15:25326432-25326525
SNORD116-16	100033428	chr:15:25327913-25328006
SNORD116-18	100033430	chr:15:25330530-25330623
SNORD116-19	727708	chr:15:25331672-25331765
SNORD116-2	100033414	chr:15:25299355-25299451
SNORD116-20	100033431	chr:15:25332807-25332900
SNORD116-21	100033432	chr:15:25333949-25334042
SNORD116-22	100033433	chr:15:25335068-25335161
SNORD116-23	100033434	chr:15:25336931-25337024
SNORD116-24	100033435	chr:15:25339182-25339275
SNORD116-25	100033436	chr:15:25342808-25342901
SNORD116-26	100033438	chr:15:25344644-25344741
SNORD116-27	100033439	chr:15:25346720-25346813
SNORD116-28	100033820	chr:15:25349787-25349879
SNORD116-29	100033821	chr:15:25351666-25351750
SNORD116-4	100033416	chr:15:25304683-25304780
SNORD116-6	100033418	chr:15:25310171-25310268
SNORD116-7	100033419	chr:15:25312933-25313029
SNORD116-8	100033420	chr:15:25315577-25315673
SNORD116-9	100033421	chr:15:25318252-25318348
SNORD64	347686	chr:15:25230246-25230312
SNRPN	6638	chr:15:25068793-25664608
SNURF	8926	chr:15:25200069-25223728
UBE3A	7337	chr:15:25582395-25684127

Table S18: Genes located in the Angelman syndrome interval.

S4.10 Chromosome 1q21.1 deletion syndrome [MIM:274000]

Also known as Thrombocytopenia-absent radius syndrome.

Term name	Term ID	Frequency	Ref.
bilateral absence of radius	HP:0004977	100.00 %	26
Aplasia/Hypoplasia of the ulna	HP:0006495	90.00 %	26
Coxa valga	HP:0002673	75.00 %	26
Dislocated hips	HP:0002827	75.00 %	26
Cow milk allergy	HP:0100327	75.00 %	26
Clinodactyly of the 5th finger	HP:0004209	75.00 %	26
Genu varum	HP:0002970	75.00 %	26
Patellar aplasia	HP:0006443	75.00 %	26
Eosinophilia	HP:0001880	75.00 %	27
Thrombocytopenia	HP:0001873	66.00 %	26,27
Leukocytosis	HP:0001974	62.00 %	27
Broad phalanges of the thumb	HP:0009651	50.00 %	26
Adducted thumbs	HP:0001181	50.00 %	26
Aplasia/Hypoplasia of the humerus	HP:0006507	50.00 %	26,27
Death in infancy	HP:0001522	40.00 %	1,27
Anemia	HP:0001903	33.00 %	27
Abnormality of the cardiac septa	HP:0001671	18.50 %	26
Tibial torsion	HP:0100694	7.50 %	27
Strabismus	HP:0000486	7.50 %	27
Ptosis	HP:0000508	7.50 %	27
Developmental delay	HP:0001263	7.50 %	27
Cerebellar hypoplasia	HP:0001321	7.50 %	27
Carpal bone hypoplasia	HP:0001498	7.50 %	27
Tetralogy of Fallot	HP:0001636	7.50 %	27
Delayed myelination	HP:0002188	7.50 %	27
Cavum septum pellucidum	HP:0002389	7.50 %	27
Dislocation of patella	HP:0002999	7.50 %	27
Nevus flammeus of the forehead	HP:0007413	7.50 %	27
Hypoplastic/small phalanges of the hand	HP:0009803	7.50 %	27
Abnormality of the shoulder	HP:0003043	7.50 %	26,27
Finger syndactyly	HP:0006101	7.50 %	26
Phocomelia	HP:0009829	7.50 %	26,27
Hepatosplenomegaly	HP:0001433	7.50 %	27
Malar hypoplasia	HP:0000272	7.50 %	27
Talipes equinovarus	HP:0001762	7.50 %	27
Edema of the dorsum of hands and feet	HP:0007514	7.50 %	27
Hooked clavicles	HP:0000895	7.50 %	27
Short stature	HP:0004322	7.00 %	27
Coarctation of aorta	HP:0001680	5.00 %	1
Axial malrotation of the kidney	HP:0004717	5.00 %	1
Cervical ribs	HP:0000891	5.00 %	26
Fused cervical vertebrae	HP:0002949	5.00 %	26
Aplasia of the uterus	HP:0000151	5.00 %	26
Fibular aplasia	HP:0002990	5.00 %	27

Term name	Term ID	Frequency	Ref.
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Table **S19**: Phenotypic annotations for the Chromosome 1q21.1 deletion syndrome [MIM:274000].

Gene Symbol	Entrez ID	Position
ANKRD34A	284615	chr:1:145470507-145475646
ANKRD35	148741	chr:1:145549208-145568525
CD160	11126	chr:1:145695797-145715564
GPR89A	653519	chr:1:145764594-145827102
GPR89C	728932	chr:1:145883867-145924048
HFE2	148738	chr:1:145413190-145417544
ITGA10	8515	chr:1:145524989-145543867
LIX1L	128077	chr:1:145477084-145499090
NUDT17	200035	chr:1:145586490-145589434
PDZK1	5174	chr:1:145727725-145764072
PEX11B	8799	chr:1:145516164-145523731
PIAS3	10401	chr:1:145575987-145586545
POLR3C	10623	chr:1:145592604-145610883
POLR3GL	84265	chr:1:145456235-145470386
RBM8A	9939	chr:1:145507637-145511443
RNF115	27246	chr:1:145611035-145688775
TXNIP	10628	chr:1:145438461-145442634

Table **S20**: Genes located in the Chromosome 1q21.1 deletion syndrome [MIM:274000] interval.

S4.11 Potocki Lupski syndrome [MIM:610883]

Term name	Term ID	Frequency	Ref.
Broad forehead	HP:0000337	100.00 %	28
Downward slanting palpebral fissures	HP:0000494	100.00 %	28
Prominent nasal tip	HP:0005274	100.00 %	28
Triangular facies	HP:0000325	100.00 %	28
Prominent jaw	HP:0002051	100.00 %	28
Micrognathia	HP:0000347	100.00 %	28
Sporadic	HP:0003745	100.00 %	
Autosomal dominant inheritance	HP:0000006	100.00 %	
Language impairment	HP:0002463	95.00 %	28
Feeding difficulties	HP:0002022	94.74 %	28
Speech articulation difficulties	HP:0009088	94.74 %	28
Generalized hypotonia	HP:0001290	90.48 %	28
Dysphasia	HP:0002357	90.00 %	28
Sleep apnea	HP:0010535	88.89 %	28

Term name	Term ID	Frequency	Ref.
Echolalia	HP:0010529	85.71 %	28
Autism	HP:0000717	83.33 %	28
EEG abnormalities	HP:0002353	78.57 %	28
Gastroesophageal reflux	HP:0002020	73.33 %	28
Failure to thrive	HP:0001508	72.22 %	28
Hypermetropia	HP:0000540	60.00 %	28
Stereotypical motor behaviors	HP:0008758	57.14 %	28
Abnormality of the cardiovascular system	HP:0001626	54.55 %	28
Low birth weight	HP:0001518	47.62 %	28
Poor eye contact	HP:0000817	42.86 %	28
Scoliosis	HP:0002650	30.77 %	28
Hypocholesterolemia	HP:0003146	30.00 %	28
Short stature	HP:0004322	23.81 %	28
Microcephaly	HP:0000252	16.67 %	28
Hypertelorism	HP:0000316	16.67 %	28
Kidney malformation	HP:0000792	15.38 %	28
Hearing impairment	HP:0000365	6.25 %	28
Epilepsy	HP:0001275	4.76 %	28

Table S21: Phenotypic annotations for Potocki Lupski syndrome [MIM:610883].

Gene Symbol	Entrez ID	Position
TNFRSF13B	23495	chr:17:16842397-16875401
FLCN	201163	chr:17:17115525-17140501
COPS3	8533	chr:17:17150138-17184590
NT5M	56953	chr:17:17206679-17250976
MED9	55090	chr:17:17380299-17396533
RASD1	51655	chr:17:17397750-17399706
PEMT	10400	chr:17:17408876-17494993
RAII	10743	chr:17:17584786-17714766
SREBF1	6720	chr:17:17714662-17740324
TOM1L2	146691	chr:17:17746821-17875783
LRRC48	83450	chr:17:17876126-17920202
ATPAF2	91647	chr:17:17921333-17942479
C17orf39	79018	chr:17:17942610-17971717
DRG2	1819	chr:17:17991282-18011291
MYO15A	51168	chr:17:18012019-18083115
ALKBH5	54890	chr:17:18086866-18113267
LLGL1	3996	chr:17:18128935-18148188
FLII	2314	chr:17:18148149-18162054
SMCR7	125170	chr:17:18163847-18169094
TOP3A	7156	chr:17:18177234-18218320
SMCR8	140775	chr:17:18218593-18231369
SHMT1	6470	chr:17:18231186-18266855
TBC1D28	254272	chr:17:18538841-18547739
TRIM16L	147166	chr:17:18625401-18639430
FBXW10	10517	chr:17:18647325-18682661

Gene Symbol	Entrez ID	Position
FAM18B1	51030	chr:17:18684581-18710025
PRPSAP2	5636	chr:17:18761491-18834579
SLC5A10	125206	chr:17:18855477-18924002
FAM83G	644815	chr:17:18874380-18908059
GRAP	10750	chr:17:18923989-18950335
EPN2	22905	chr:17:19140689-19240027
B9D1	27077	chr:17:19246482-19266045
MAPK7	5598	chr:17:19281033-19286856
MFAP4	4239	chr:17:19286754-19290492
SLC47A1	55244	chr:17:19437166-19482345
ALDH3A2	224	chr:17:19552063-19580907
SLC47A2	146802	chr:17:19581627-19620042
ALDH3A1	218	chr:17:19641296-19651745
ULK2	9706	chr:17:19674142-19771238
AKAP10	11216	chr:17:19808748-19881128
SPECC1	92521	chr:17:19990334-20218067
SMCR5	140771	chr:17:17679999-17682842
SNORA59B	677882	chr:17:19460872-19461023
SNORD3B-1	26851	chr:17:18965224-18965440

Table **S22:** Genes located in the Potocki Lupski syndrome [MIM:610883] interval.

S4.12 Renal Cysts and Diabetes (RCAD) syndrome [MIM:137920]

Term name	Term ID	Frequency	Ref.
Autosomal dominant inheritance	HP:0000006	100.00 %	1
Variable age at onset	HP:0003618	100.00 %	1
Phenotypic variability	HP:0003812	100.00 %	1
Exocrine pancreatic insufficiency	HP:0001738	85.71 %	29
Pancreatic hypoplasia	HP:0002594	83.33 %	29
Renal cysts	HP:0000107	82.61 %	30
Insulin-dependent maturity-onset diabetes of the young	HP:0004904	76.92 %	29
Abnormality of alkaline phosphatase activity	HP:0004379	57.14 %	30
Proteinuria	HP:0000093	33.00 %	30
Chronic renal failure	HP:0000101	33.00 %	MIM:137920
Glucose intolerance	HP:0000833	33.00 %	30
Gout	HP:0001997	33.00 %	30
Glycosuria	HP:0003076	33.00 %	1
Increased creatinine	HP:0003259	33.00 %	1
Abnormal liver function tests	HP:0001411	33.00 %	30
Biliary tract abnormality	HP:0001080	33.00 %	30
Hypoplastic glomerulocystic kidney disease	HP:0100611	17.39 %	30
Kidney malformation	HP:0000792	8.70 %	30

Term name	Term ID	Frequency	Ref.
Diabetes mellitus	HP:0000819	7.50 %	30
Renal hypoplasia	HP:0000089	4.35 %	30
Unilateral renal agenesis	HP:0000122	4.35 %	30
Bicornuate uterus	HP:0000813	4.35 %	30
Hypoplasia of the uterus	HP:0000013	4.35 %	30

Table **S23**: Phenotypic annotations for Renal Cysts and Diabetes (RCAD) syndrome [MIM:137920].

Gene Symbol	Entrez ID	Position
AATF	26574	chr:17:35306174-35414170
ACACA	31	chr:17:35441926-35766901
C17orf78	284099	chr:17:35732984-35749661
DDX52	11056	chr:17:35972412-36003486
DUSP14	11072	chr:17:35849950-35873587
GGNBP2	79893	chr:17:34900736-34946277
HNF1B	6928	chr:17:36046433-36105095
LHX1	3975	chr:17:35294498-35300493
MRM1	79922	chr:17:34958024-34965406
SYNRG	11276	chr:17:35874899-35969485
TADA2A	6871	chr:17:35766976-35837225

Table **S24**: Genes located in the Renal Cysts and Diabetes (RCAD) syndrome [MIM:137920] interval.

S4.13 Leri Weill Dyschondrosteosis [MIM:127300]

Term name	Term ID	Frequency	Reference
Autosomal dominant inheritance	HP:0000006	100.00 %	1
Mesomelia	HP:0003027	90.00 %	31
Short stature, disproportionate short-limbed	HP:0008873	90.00 %	31
Madelung deformity	HP:0003067	80.00 %	31
Madelung deformity	HP:0003067	63.50 %	31
Hypoplastic tibia	HP:0005736	50.00 %	2
Limited elbow movement	HP:0002996	50.00 %	1
Hypoplasia of the radius	HP:0002984	50.00 %	31
Limited wrist movement	HP:0006248	50.00 %	1
Radial bowing	HP:0002986	50.00 %	31
Hypoplasia of the ulna	HP:0003022	50.00 %	31
Dorsal subluxation of ulna	HP:0006459	50.00 %	31
Abnormality of the carpal bones	HP:0001191	50.00 %	31
Fibular hypoplasia	HP:0003038	50.00 %	2
High-arched palate	HP:0000156	7.50 %	31,32
Scoliosis	HP:0002650	7.50 %	31,32
Increased carrying angle	HP:0003102	7.50 %	31
Muscle hypertrophy	HP:0003712	7.50 %	31
Hypoplastic/short 4th metacarpal	HP:0010044	7.50 %	31
Multiple exostoses	HP:0002762	7.50 %	31,32
Brachydactyly (feet)	HP:0001831	7.50 %	2
Abnormality of the metatarsal bones	HP:0001832	7.50 %	2
Tibial bowing	HP:0002982	7.50 %	2
Abnormality of the femoral neck	HP:0003367	7.50 %	2
Coxa valga	HP:0002673	7.50 %	2
Abnormality of the humerus	HP:0003063	7.50 %	2

Table S25: Phenotypic annotations for Leri Weill Dyschondrosteosis [MIM:127300].

Gene Symbol	Entrez ID	Position
SHOX	6473	chr:X:585078-620145

Table S26: Gene located in the Leri Weill Dyschondrosteosis [MIM:127300] interval.

S4.14 Prader Willi Syndrome [MIM:176270]

Term name	Term ID	Frequency	Ref.
Dolichocephaly	HP:0000268	100.00 %	1
Hypermetropia	HP:0000540	100.00 %	1
Hyperinsulinemia	HP:0000842	100.00 %	1
Isolated cases	HP:0001420	100.00 %	1
Hypoventilation	HP:0002791	100.00 %	1
Decreased fetal movement	HP:0001558	100.00 %	33
Hypogonadotrophic hypogonadism	HP:0000044	90.00 %	33
Growth hormone deficiency	HP:0000824	90.00 %	33
Hypoplastic hand	HP:0004279	90.00 %	33
Learning disability	HP:0001328	90.00 %	33
Failure to thrive in infancy	HP:0001531	90.00 %	33
Small feet	HP:0001764	90.00 %	33
Poor suck	HP:0002033	90.00 %	33
Generalized hypotonia	HP:0001290	90.00 %	33
Feeding problems in infancy	HP:0008872	90.00 %	33
Motor delay	HP:0001270	90.00 %	33
Impaired language development	HP:0000750	90.00 %	33
Infertility	HP:0000789	90.00 %	33
Hyperphagia	HP:0000724	90.00 %	33
Obesity	HP:0001513	90.00 %	33
Short stature	HP:0004322	90.00 %	33
Narrow hand	HP:0004283	90.00 %	33
Cryptorchidism	HP:0000028	85.00 %	33
Attention deficit hyperactivity disorder	HP:0007018	75.00 %	33
Scrotal hypoplasia	HP:0000046	69.00 %	33
Adrenal insufficiency	HP:0000846	60.00 %	33
Primary amenorrhea	HP:0000786	56.00 %	33
Micropenis	HP:0000054	50.00 %	33
Clitoral hypoplasia	HP:0000060	50.00 %	33
Hypoplastic labia minora	HP:0000064	50.00 %	33
Thin upper lip vermillion	HP:0000219	50.00 %	33
Bitemporal narrowing	HP:0000314	50.00 %	33
Photosensitivity	HP:0000992	50.00 %	2
Hypernasal speech	HP:0001614	50.00 %	2
Decreased muscle mass	HP:0003199	50.00 %	33
Delayed puberty	HP:0000823	50.00 %	33
Downturned corners of mouth	HP:0002714	50.00 %	33
Almond-shaped palpebral fissures	HP:0007874	50.00 %	33
Narrow nasal bridge	HP:0000446	50.00 %	33
Scoliosis	HP:0002650	50.00 %	33
Recurrent respiratory infections	HP:0002205	50.00 %	33
Kyphosis	HP:0002808	50.00 %	33
Sleep apnea	HP:0010535	50.00 %	33
Oligomenorrhea	HP:0000876	33.00 %	33
Hypopigmentation of the skin	HP:0001010	33.00 %	33
Hypopigmentation of hair	HP:0005599	33.00 %	33
Reduced iris pigmentation	HP:0007730	33.00 %	33
Ventriculomegaly	HP:0002119	33.00 %	33
Decreased pain sensation	HP:0007328	33.00 %	33

Term name	Term ID	Frequency	Ref.
Noninsulin-dependent diabetes mellitus	HP:0005978	25.00 %	33
Autism	HP:0000717	19.00 %	33
Seizures	HP:0001250	15.00 %	33
Psychosis	HP:0000709	15.00 %	33
Hip dysplasia	HP:0001385	10.00 %	33
Myopia	HP:0000545	7.50 %	33
Upplanting palpebral fissures	HP:0000582	7.50 %	2
Osteopenia	HP:0000938	7.50 %	33
Osteoporosis	HP:0000939	7.50 %	33
Radial deviation of fingers	HP:0009466	7.50 %	2
Syndactyly	HP:0001159	7.50 %	2
Frontal hair upsweep	HP:0002236	7.50 %	2
Carious teeth	HP:0000670	7.50 %	2
Poor fine motor coordination	HP:0007010	7.50 %	2
Esotropia	HP:0000565	7.50 %	33
Temperature instability	HP:0005968	7.50 %	2
Precocious puberty	HP:0000826	4.00 %	33

Table S27: Phenotypic annotations for the Prader Willi Syndrome
[MIM:176270]

Gene Symbol	Entrez ID	Position
ATP10A	57194	chr:15:25923858-26108348
C15orf2	23742	chr:15:24920540-24928592
CYFIP1	23191	chr:15:22892683-23003602
GABRA5	2558	chr:15:27111865-27194356
GABRB3	2562	chr:15:26788692-27018934
GABRG3	2567	chr:15:27216428-27778372
HERC2	8924	chr:15:28356185-28567294
MKRN3	7681	chr:15:23810453-23813166
NDN	4692	chr:15:23930553-23932449
NIPA1	123606	chr:15:23043278-23086842
NIPA2	81614	chr:15:23004683-23034426
OCA2	4948	chr:15:28000020-28344457
SNORD107	91380	chr:15:25227140-25227214
SNORD109B	338429	chr:15:25523489-25523555
SNORD115-1	338433	chr:15:25415869-25415950
SNORD116-1	100033413	chr:15:25296622-25296718
SNORD116-10	100033422	chr:15:25319259-25319362
SNORD116-11	100033423	chr:15:25321074-25321167
SNORD116-12	100033424	chr:15:25322196-25322289
SNORD116-13	100033425	chr:15:25324203-25324296
SNORD116-14	100033426	chr:15:25325287-25325380
SNORD116-15	100033427	chr:15:25326432-25326525
SNORD116-16	100033428	chr:15:25327913-25328006
SNORD116-18	100033430	chr:15:25330530-25330623
SNORD116-19	727708	chr:15:25331672-25331765
SNORD116-2	100033414	chr:15:25299355-25299451

Gene Symbol	Entrez ID	Position
SNORD116-20	100033431	chr:15:25332807-25332900
SNORD116-21	100033432	chr:15:25333949-25334042
SNORD116-22	100033433	chr:15:25335068-25335161
SNORD116-23	100033434	chr:15:25336931-25337024
SNORD116-24	100033435	chr:15:25339182-25339275
SNORD116-25	100033436	chr:15:25342808-25342901
SNORD116-26	100033438	chr:15:25344644-25344741
SNORD116-27	100033439	chr:15:25346720-25346813
SNORD116-28	100033820	chr:15:25349787-25349879
SNORD116-29	100033821	chr:15:25351666-25351750
SNORD116-4	100033416	chr:15:25304683-25304780
SNORD116-6	100033418	chr:15:25310171-25310268
SNORD116-7	100033419	chr:15:25312933-25313029
SNORD116-8	100033420	chr:15:25315577-25315673
SNORD116-9	100033421	chr:15:25318252-25318348
SNORD64	347686	chr:15:25230246-25230312
SNRPN	6638	chr:15:25068793-25664608
SNURF	8926	chr:15:25200069-25223728
UBE3A	7337	chr:15:25582395-25684127

Table **S28:** Genes located in the Prader Willi Syndrome [MIM:176270] interval.

S4.15 Split hand/foot malformation 1 [MIM:183600]

Term name	Term ID	Frequency	Ref.
Autosomal dominant inheritance	HP:0000006	100.00 %	³⁴
Ectrodactyly (feet)	HP:0001839	90.00 %	¹
Ectrodactyly (hands)	HP:0001171	90.00 %	¹
Hearing impairment	HP:0000365	35.00 %	³⁴
Abnormality of the pinna	HP:0000377	35.00 %	³⁴
Oligodactyly (hands)	HP:0001180	33.00 %	³⁴
Intellectual disability	HP:0001249	33.00 %	³⁴
Cleft palate	HP:0000175	33.00 %	¹
Oligodactyly (feet)	HP:0001849	33.00 %	³⁴
Syndactyly	HP:0001159	33.00 %	³⁴
Cardiac malformation	HP:0002564	13.00 %	³⁴

Table S29: Phenotypic annotations for Split hand/foot malformation 1 [MIM:183600].

Gene Symbol	Entrez ID	Position
ACN9	57001	chr:7:96745904-96811074
DLX5	1749	chr:7:96649701-96654142
DLX6	1750	chr:7:96635289-96640351
DYNC1I1	1780	chr:7:95401817-95727735
SHFM1	7979	chr:7:96318078-96339202
SLC25A13	10165	chr:7:95749531-95951458

Table S30: Genes located in the Split hand/foot malformation 1 [MIM:183600] interval.

S4.16 Rubinstein Taybi Syndrome [MIM:180849]

Term name	Term ID	Frequency	Ref.
Autosomal dominant inheritance	HP:0000006	100.00 %	³⁵
High-arched palate	HP:0000156	100.00 %	^{2,35}
Hypoplasia of the maxilla	HP:0000327	100.00 %	²
Intellectual disability	HP:0001249	100.00 %	^{2,35}
Broad hallux	HP:0010055	100.00 %	²
Heterogeneous	HP:0001425	100.00 %	³⁵
Sporadic	HP:0003745	100.00 %	³⁵
Beaked nose	HP:0000444	90.00 %	²
Columella, low hanging	HP:0009765	90.00 %	^{2,35}
Speech difficulties	HP:0006936	90.00 %	²
Downward slanting palpebral fissures	HP:0000494	88.00 %	²
Long eyelashes	HP:0000527	87.00 %	²
Broad phalanges of the thumb	HP:0009651	87.00 %	²
Radial deviation of thumb terminal phalanx	HP:0005895	87.00 %	²
Unsteady gait	HP:0002317	85.00 %	²
Low-set ears	HP:0000369	84.00 %	²
Abnormality of the pinna	HP:0000377	84.00 %	²
Feeding problems in infancy	HP:0008872	80.00 %	^{35,36}
Cryptorchidism	HP:0000028	78.00 %	²
Thick eyebrows	HP:0000574	76.00 %	²
Hirsutism	HP:0001007	75.00 %	²
Poor coordination	HP:0002370	75.00 %	²
Delayed skeletal maturation	HP:0002750	74.00 %	²
Highly arched eyebrows	HP:0001584	73.00 %	²
Pes planus	HP:0001763	72.00 %	²
Deviated nasal septum	HP:0004411	71.00 %	²
Strabismus	HP:0000486	69.00 %	²
Muscular hypotonia	HP:0001252	67.00 %	²
Clinodactyly of the 5th finger	HP:0004209	62.00 %	²
Recurrent upper respiratory tract infections	HP:0002788	60.00 %	^{35,36}
EEG abnormalities	HP:0002353	57.00 %	²
Small mouth	HP:0000160	56.00 %	²
Epicanthus	HP:0000286	55.00 %	²
Abnormality of the kidney	HP:0000077	52.00 %	²
Respiratory difficulties	HP:0002880	51.00 %	^{35,36}
Short stature	HP:0004322	50.00 %	³⁵
Constipation	HP:0002019	50.00 %	³⁵
Facial grimacing	HP:0000273	50.00 %	³⁵
Talon cusp	HP:0011087	50.00 %	³⁵
Poor weight gain	HP:0001535	50.00 %	³⁵
Micrognathia	HP:0000347	49.00 %	²
Spina bifida occulta	HP:0003298	47.00 %	²
Nasolacrimal duct obstruction	HP:0000579	43.00 %	²
Low posterior hairline	HP:0002162	42.00 %	²
Scoliosis	HP:0002650	42.00 %	²
Wide anterior fontanel	HP:0000260	41.00 %	²
Abnormality of refraction	HP:0000539	41.00 %	^{2,35}

Term name	Term ID	Frequency	Ref.
Hyperreflexia	HP:0001347	40.00 %	2
Abnormality of the cervical spine	HP:0003319	37.00 %	2
Ptosis	HP:0000508	36.00 %	2
Microcephaly	HP:0000252	35.00 %	2
Frontal bossing	HP:0002007	33.00 %	2
Short attention span	HP:0000736	33.00 %	35
Ventricular septal defect	HP:0001629	33.00 %	2
Atrial septal defect	HP:0001631	33.00 %	2
Patent ductus arteriosus	HP:0001643	33.00 %	2
Plantar crease between first and second toes	HP:0008107	33.00 %	2
Truncal obesity	HP:0001956	33.00 %	35
Obstructive sleep apnea	HP:0002870	33.00 %	35
Laryngomalacia	HP:0001601	33.00 %	35
Impulsivity	HP:0100710	33.00 %	35
Autism	HP:0000717	33.00 %	35
Phonophobia	HP:0002183	33.00 %	35
Agoraphobia	HP:0000756	33.00 %	35
Chorioretinal dystrophy	HP:0001135	31.00 %	
Prominent fingertip pads	HP:0001212	31.00 %	2
Polyhydramnios	HP:0001561	30.00 %	36
Small, flared iliac wings	HP:0003181	26.00 %	2
Capillary hemangiomas	HP:0005306	25.00 %	2
Delayed closure of fontanelles	HP:0000270	24.00 %	2
Low anterior hairline	HP:0000294	24.00 %	2
Hearing impairment	HP:0000365	24.00 %	36
Seizures	HP:0001250	23.00 %	2
Enophthalmos	HP:0000663	22.00 %	2
Keloids	HP:0010562	22.00 %	2
Frontal hair upsweep	HP:0002236	20.00 %	2
Hypospadias	HP:0000047	8.00 %	2
Shawl scrotum	HP:0000049	8.00 %	2
Glaucoma	HP:0000501	8.00 %	2
Cataract	HP:0000518	8.00 %	2
Coloboma	HP:0000589	8.00 %	2
Parietal foramina	HP:0002697	8.00 %	2
Large foramen magnum	HP:0002700	8.00 %	2
Dislocation of patella	HP:0002999	8.00 %	2,35
Duane anomaly	HP:0009921	8.00 %	2
Proptosis	HP:0000520	8.00 %	2
Abnormality of cardiac conduction	HP:0001665	8.00 %	2
Contractures	HP:0001371	8.00 %	2
Transverse palmar creases	HP:0000954	8.00 %	2
High axial triradius	HP:0001042	8.00 %	2
Partial/complete duplication of the phalanges of the hallux	HP:0010066	8.00 %	2,35
Dislocated radial head	HP:0003083	8.00 %	2
Avascular necrosis of the capital femoral epiphysis	HP:0005743	8.00 %	2,35
Bifid uterus	HP:0000136	8.00 %	2
Papillary cystadenoma of the epididymis	HP:0009715	8.00 %	2

Term name	Term ID	Frequency	Ref.
Pectus excavatum	HP:0000767	8.00 %	2
Congenital megacolon	HP:0002251	8.00 %	2
Stereotypical motor behaviors	HP:0008758	8.00 %	2
Mirror hand movements (bimanual synkinesia)	HP:0001335	8.00 %	2
Tethered cord	HP:0002144	8.00 %	2
Vascular ring	HP:0010775	8.00 %	2
Premature thelarche	HP:0010314	8.00 %	2
Broad nasal bridge	HP:0000431	7.50 %	37
Syndactyly	HP:0001159	7.50 %	2
Agenesis of corpus callosum	HP:0001274	7.50 %	2
Abnormality of the cornea	HP:0000481	7.50 %	35
Joint laxity	HP:0001388	7.50 %	35
Dental malocclusion	HP:0000689	7.50 %	35
Dental crowding	HP:0000678	7.50 %	35
Abnormal number of teeth	HP:0006483	7.50 %	35
Hyperactivity	HP:0000752	7.50 %	35
Self-mutilation	HP:0000742	7.50 %	35

Table **S31**: Phenotypic annotations for the Rubinstein Taybi Syndrome [MIM:180849].

Gene Symbol	Entrez ID	Position
CREBBP	1387	chr:16:3775054-3930120

Table **S32**: Gene located in the interval of the Rubinstein Taybi Syndrome [MIM:180849].

S4.17 Chromosome 22q13.3 Deletion Syndrome [MIM:606232]

Also known as Phelan-McDermid Syndrome.

Term name	Term ID	Frequency	Ref.
Sporadic	HP:0003745	100.00 %	38
Impaired language development	HP:0000750	90.00 %	38
Neonatal hypotonia	HP:0001319	90.00 %	38
Normal to tall stature	HP:0003516	90.00 %	38
Large hands	HP:0001176	75.00 %	38
Hypoplastic toenails	HP:0001800	75.00 %	38
Long eyelashes	HP:0000527	75.00 %	38
Decreased pain sensation	HP:0007328	75.00 %	38
Bruxism	HP:0003763	75.00 %	38
Hyperorality	HP:0000710	75.00 %	38
Poor eye contact	HP:0000817	75.00 %	39
Stereotypical motor behaviors	HP:0008758	75.00 %	39
Autism	HP:0000717	75.00 %	38
Dolichocephaly	HP:0000268	50.00 %	38
Large, prominent ears	HP:0000382	50.00 %	38
Thick eyebrows	HP:0000574	50.00 %	38
Full cheeks	HP:0000293	50.00 %	38
Palpebral edema	HP:0100540	50.00 %	38
Deeply set eye	HP:0000490	50.00 %	38
Malar hypoplasia	HP:0000272	50.00 %	38
Broad nasal bridge	HP:0000431	50.00 %	38
Bulbous nose	HP:0000414	50.00 %	38
Pointed chin	HP:0000307	50.00 %	38
Sacral dimple	HP:0000960	50.00 %	38
Hypohidrosis	HP:0000966	50.00 %	38
Heat intolerance	HP:0002046	50.00 %	38
Ptosis	HP:0000508	50.00 %	39
Broad-based gait	HP:0002136	50.00 %	38
Unsteady gait	HP:0002317	50.00 %	38
Strabismus	HP:0000486	33.00 %	38
Gastroesophageal reflux	HP:0002020	33.00 %	38
Dental malocclusion	HP:0000689	33.00 %	38
Epicanthus	HP:0000286	33.00 %	38
Long philtrum	HP:0000343	33.00 %	38
High-arched palate	HP:0000156	33.00 %	38
Episodic vomiting	HP:0002572	33.00 %	39
2-3 toe syndactyly	HP:0004691	33.00 %	39
Clinodactyly of the 5th finger	HP:0004209	33.00 %	39
Seizures	HP:0001250	33.00 %	39
Lymphedema	HP:0001004	33.00 %	39
Aggressive behavior	HP:0000718	25.00 %	38
Hearing impairment	HP:0000365	20.00 %	39
Arachnoid cyst	HP:0100702	15.00 %	39
Tongue thrusting	HP:0100703	15.00 %	39
Cellulitis	HP:0100658	10.00 %	39
Ventriculomegaly	HP:0002119	7.50 %	2
Delayed myelination	HP:0002188	7.50 %	2

Term name	Term ID	Frequency	Ref.
Abnormality of the periventricular white matter	HP:0002518	7.50 %	2
Patent ductus arteriosus	HP:0001643	7.50 %	2
Ventricular septal defect	HP:0001629	7.50 %	2
Vesicoureteral reflux	HP:0000076	7.50 %	2
Polycystic kidney dysplasia	HP:0000113	7.50 %	2
Cortical visual impairment	HP:0100704	6.00 %	38
Microcephaly	HP:0000252	5.00 %	38

Table **S33**: Phenotypic annotations for the Chromosome 22q13.3 Deletion Syndrome [MIM:606232].

Gene Symbol	Entrez ID	Position
ACR	49	chr:22:51176651-51183726
ARSA	410	chr:22:51063448-51066606
MAPK8IP2	23542	chr:22:51039130-51049978
SHANK3	85358	chr:22:51113069-51171640

Table **S34**: Genes located in the interval for the Chromosome 22q13.3 Deletion Syndrome [MIM:606232].

S4.18 Pelizaeus Merzbacher Disease [MIM:312080]

Term name	Term ID	Frequency	Ref.
Microcephaly	HP:0000252	100.00 %	¹
X-linked recessive inheritance	HP:0001419	100.00 %	⁴⁰
Nystagmus	HP:0000639	100.00 %	⁴¹
Psychomotor degeneration	HP:0002361	100.00 %	⁴¹
Infantile onset	HP:0003593	100.00 %	⁴⁰
Ataxia	HP:0001251	90.00 %	⁴¹
Muscular hypotonia	HP:0001252	90.00 %	⁴⁰
Progressive spastic quadriplegia	HP:0002478	90.00 %	⁴⁰
Dystonia	HP:0001332	90.00 %	⁴¹
Slow progression	HP:0003677	90.00 %	⁴¹
Developmental delay	HP:0001263	90.00 %	⁴²
Pyramidal signs	HP:0007256	90.00 %	⁴¹
Abnormal myelination	HP:0002520	90.00 %	⁴¹
Dysarthria	HP:0001260	75.00 %	⁴⁰
Choreoathetosis	HP:0001266	75.00 %	⁴⁰
Sudanophilic leukodystrophy	HP:0003269	75.00 %	⁴¹
Reduction of oligodendroglia	HP:0100709	75.00 %	^{40,43}
Dysphagia	HP:0002015	75.00 %	⁴²
Hyporeflexia	HP:0001265	57.14 %	⁴¹
Impaired language development	HP:0000750	50.00 %	⁴⁰
Head titubation	HP:0002599	50.00 %	⁴¹
Short stature	HP:0004322	50.00 %	⁴⁰
Poor weight gain	HP:0001535	50.00 %	⁴⁰
Optic atrophy	HP:0000648	33.00 %	⁴¹
Seizures	HP:0001250	7.50 %	⁴²
Congenital laryngeal stridor	HP:0004886	7.50 %	^{41,42}

Table S35: Phenotypic annotations for Pelizaeus Merzbacher Disease [MIM:312080].

Gene Symbol	Entrez ID	Position
GLRA4	441509	chr:X:102962271-102983551
MORF4L2	9643	chr:X:102930425-102943085
PLP1	5354	chr:X:103031438-103047547
RAB40A	142684	chr:X:102754680-102774416
RAB9B	51209	chr:X:103077254-103087211
TCEAL1	9338	chr:X:102883647-102885880
TCEAL3	85012	chr:X:102862833-102864854
TCEAL4	79921	chr:X:102840418-102842656
TMEM31	203562	chr:X:102965836-102968959

Table S36: Genes located in the interval for Pelizaeus Merzbacher Disease [MIM:312080].

S4.19 Sotos syndrome [MIM:117550]

Term name	Term ID	Frequency	Ref.
Isolated cases	HP:0001420	100.00 %	¹
Autosomal dominant inheritance	HP:0000006	100.00 %	⁴⁴
Accelerated skeletal maturation	HP:0005616	84.00 %	²
Macrocephaly	HP:0000256	75.00 %	²
High-arched palate	HP:0000156	50.00 %	²
Frontal bossing	HP:0002007	50.00 %	²
Dolichocephaly	HP:0000268	50.00 %	²
Pointed chin	HP:0000307	50.00 %	⁴⁴
Downward slanting palpebral fissures	HP:0000494	50.00 %	²
Large hands	HP:0001176	50.00 %	²
Neonatal hypotonia	HP:0001319	50.00 %	⁴⁴
Joint laxity	HP:0001388	50.00 %	⁴⁴
Large feet	HP:0001833	50.00 %	²
Expressive language delay	HP:0002474	50.00 %	⁴⁴
Advanced eruption of teeth	HP:0006288	50.00 %	²
Cavum septum pellucidum	HP:0002389	50.00 %	⁴⁴
Partial agenesis of the corpus callosum	HP:0001338	50.00 %	⁴⁴
High anterior hairline	HP:0009890	50.00 %	²
Tall stature	HP:0000098	50.00 %	²
Prominent jaw	HP:0002051	50.00 %	²
Poor coordination	HP:0002370	50.00 %	⁴⁴
Scoliosis	HP:0002650	30.00 %	⁴⁴
Seizures	HP:0001250	25.00 %	⁴⁴
Abnormal glucose tolerance	HP:0001952	14.00 %	²
Otitis media	HP:0000388	7.50 %	⁴⁴
Conductive hearing impairment	HP:0000405	7.50 %	⁴⁴
Strabismus	HP:0000486	7.50 %	⁴⁴
Hypermetropia	HP:0000540	7.50 %	⁴⁴
Nystagmus	HP:0000639	7.50 %	⁴⁴
Hyperreflexia	HP:0001347	7.50 %	¹
Nail hypoplasia	HP:0001792	7.50 %	⁴⁴
Ventricular septal defect	HP:0001629	7.50 %	⁴⁴
Atrial septal defect	HP:0001631	7.50 %	⁴⁴
Patent ductus arteriosus	HP:0001643	7.50 %	⁴⁴
Enlarged cisterna magna	HP:0002280	7.50 %	⁴⁴
Hypertelorism	HP:0000316	7.50 %	²
Pes planus	HP:0001763	7.50 %	⁴⁴
Nephroblastoma (Wilms tumor)	HP:0002667	5.00 %	⁴⁴

Table S37: Phenotypic annotations for Sotos syndrome [MIM:117550].

Gene Symbol	Entrez ID	Position
ARL10	285598	chr:5:175792501-175800502
B4GALT7	11285	chr:5:177027118-177037347

Gene Symbol	Entrez ID	Position
C5orf25	375484	chr:5:175665369-175772991
CDHR2	54825	chr:5:175969511-176022768
CLTB	1212	chr:5:175819455-175843539
CPLX2	10814	chr:5:175223609-175311022
DBN1	1627	chr:5:176883613-176900693
DDX41	51428	chr:5:176938577-176943966
DOK3	79930	chr:5:176928904-176937426
F12	2161	chr:5:176829138-176836576
FAF2	23197	chr:5:175875355-175937074
FAM153A	285596	chr:5:177150364-177207504
FAM153B	202134	chr:5:175511908-175543457
FGFR4	2264	chr:5:176513920-176525126
GPRIN1	114787	chr:5:176022802-176037130
GRK6	2870	chr:5:176853686-176869849
HIGD2A	192286	chr:5:175815783-175816750
HK3	3101	chr:5:176307869-176326332
KIAA1191	57179	chr:5:175773064-175788808
LMAN2	10960	chr:5:176758562-176778884
MXD3	83463	chr:5:176732500-176739291
NSD1	64324	chr:5:176560079-176727213
PDLIM7	9260	chr:5:176910394-176924601
PFN3	345456	chr:5:176827107-176827636
PRELID1	27166	chr:5:176730834-176733949
PROP1	5626	chr:5:177419235-177423242
PRR7	80758	chr:5:176873795-176883286
RAB24	53917	chr:5:176728198-176730743
RGS14	10636	chr:5:176784843-176799598
RNF44	22838	chr:5:175953699-175964420
SLC34A1	6569	chr:5:176811431-176825848
SNCB	6620	chr:5:176047209-176057556
THOC3	84321	chr:5:175386533-175395544
TMED9	54732	chr:5:177019212-177023107
TSPAN17	26262	chr:5:176074387-176086058
UIMC1	51720	chr:5:176332005-176433442
ZNF346	23567	chr:5:176449696-176493757

Table **S38:** Genes located in the interval for Sotos syndrome [MIM:117550].

S4.20 Wolf Hirschhorn Syndrome [MIM:194190]

Term name	Term ID	Frequency	Reference
Absent septum pellucidum	HP:0001331	100.00 %	1
Decreased fetal movement	HP:0001558	100.00 %	1
Ventricular septal defect	HP:0001629	100.00 %	1
Accessory spleen	HP:0001747	100.00 %	1
Hyperconvex fingernails	HP:0001812	100.00 %	1
Gastroesophageal reflux	HP:0002020	100.00 %	1
Low posterior hairline	HP:0002162	100.00 %	1
Cavum septum pellucidum	HP:0002389	100.00 %	1
Delayed skeletal maturation	HP:0002750	100.00 %	1
Vertebral fusion	HP:0002948	100.00 %	1
Malrotation of small bowel	HP:0004794	100.00 %	1
Pseudoepiphyses of the metacarpals	HP:0009193	100.00 %	1
Sternal ossification center abnormalities	HP:0006624	100.00 %	1
Sporadic	HP:0003745	100.00 %	1
Autosomal dominant inheritance	HP:0000006	100.00 %	1
Low birth weight	HP:0001518	90.00 %	45
Developmental delay	HP:0001263	90.00 %	45
Intrauterine growth restriction	HP:0001511	90.00 %	27,45
Generalized hypotonia	HP:0001290	90.00 %	45
Decreased muscle mass	HP:0003199	90.00 %	45
EEG abnormalities	HP:0002353	90.00 %	45
Ptosis	HP:0000508	75.00 %	45
Seizures	HP:0001250	75.00 %	45
Failure to thrive	HP:0001508	75.00 %	27,45
Severe growth retardation	HP:0001521	75.00 %	27
Short stature	HP:0004322	75.00 %	27,45
Immunodeficiency	HP:0002721	69.00 %	45
Hypospadias	HP:0000047	50.00 %	45
Short upper lip	HP:0000188	50.00 %	1
Micrognathia	HP:0000347	50.00 %	45
Microcephaly	HP:0000252	50.00 %	45
Craniofacial asymmetry	HP:0004484	50.00 %	45
Epicanthus	HP:0000286	50.00 %	45
Hypertelorism	HP:0000316	50.00 %	45
Short philtrum	HP:0000322	50.00 %	45
High forehead	HP:0000348	50.00 %	45
Preauricular skin tag	HP:0000384	50.00 %	45
Stenotic external auditory canal	HP:0000402	50.00 %	1,27
Broad nasal bridge	HP:0000431	50.00 %	45
Beaked nose	HP:0000444	50.00 %	1,27
Strabismus	HP:0000486	50.00 %	1,27
Rieger anomaly	HP:0000558	50.00 %	1,27
Nystagmus	HP:0000639	50.00 %	1,27
Hypodontia	HP:0000668	50.00 %	1,27
Highly arched eyebrows	HP:0001584	50.00 %	45
Downturned corners of mouth	HP:0002714	50.00 %	45
Preauricular pit	HP:0004467	50.00 %	45
Prominent glabella	HP:0002057	50.00 %	45

Term name	Term ID	Frequency	Reference
Cryptorchidism	HP:0000028	50.00 %	45
Poorly formed pinnae	HP:0008562	50.00 %	45
Conductive hearing impairment	HP:0000405	40.00 %	45
Cleft lip/palate	HP:0000202	33.00 %	45
Rib fusion	HP:0000902	33.00 %	45
Scoliosis	HP:0002650	33.00 %	45
Kyphosis	HP:0002808	33.00 %	45
Stereotypical motor behaviors	HP:0008758	33.00 %	45
Abnormal form of the vertebral bodies	HP:0003312	33.00 %	45
Rib segmentation abnormalities	HP:0006655	33.00 %	45
Talipes equinovarus	HP:0001762	33.00 %	45
Malformation of the central nervous system	HP:0007319	33.00 %	45
Hemangiomas	HP:0001028	33.00 %	1,27
Proptosis	HP:0000520	33.00 %	27
Iris coloboma	HP:0000612	30.00 %	27
Atrial septal defect	HP:0001631	27.00 %	45
Genitourinary tract malformation	HP:0008713	25.00 %	45
Sensorineural hearing impairment	HP:0000407	15.00 %	45
Preaxial polydactyly (feet)	HP:0001841	7.50 %	27
Hip dysplasia	HP:0001385	7.50 %	27
Ectrodactyly (hands)	HP:0001171	7.50 %	45
Abnormality of the lacrimal duct	HP:0000614	7.50 %	27
Preaxial polydactyly (hands)	HP:0001177	7.50 %	27
Hypoplastic/small hallux	HP:0010109	7.50 %	27
Hypoplastic/small thumb	HP:0009778	7.50 %	27
Aplasia cutis congenita of scalp	HP:0007385	7.50 %	1,27
Agenesis of corpus callosum	HP:0001274	7.50 %	27
Aplasia of the uterus	HP:0000151	5.00 %	1
Hydrocephalus	HP:0000238	5.00 %	1
Webbed neck	HP:0000465	5.00 %	1
Transverse palmar creases	HP:0000954	5.00 %	1
Sacral dimple	HP:0000960	5.00 %	1
Biliary tract abnormality	HP:0001080	5.00 %	1

Table S39: Phenotypic annotations for Wolf Hirschhorn Syndrome [MIM:194190].

Gene Symbol	Entrez ID	Position
ATP5I	521	chr:4:666224-668126
C4orf42	92070	chr:4:1243227-1246794
CRIPAK	285464	chr:4:1385339-1389781
CTBP1	1487	chr:4:1205227-1242907
DGKQ	1609	chr:4:952674-967343
FAM53A	152877	chr:4:1641607-1685987
FGFR3	2261	chr:4:1795038-1810598
FGFRL1	53834	chr:4:1005609-1020685
GAK	2580	chr:4:843064-926173

Gene Symbol	Entrez ID	Position
IDUA	3425	chr:4:980784-998316
KIAA1530	57654	chr:4:1341103-1381836
LETM1	3954	chr:4:1813205-1857973
MAEA	10296	chr:4:1283671-1333924
MFSD7	84179	chr:4:675617-682972
MYL5	4636	chr:4:671710-675816
NAT8L	339983	chr:4:2061238-2070815
NKX1-1	54729	chr:4:1396719-1400118
PCGF3	10336	chr:4:699572-764427
PDE6B	5158	chr:4:619362-664680
PIGG	54872	chr:4:492988-533319
POLN	353497	chr:4:2073644-2230957
RNF212	285498	chr:4:1065265-1107581
SCARNA22	677770	chr:4:1976362-1976486
SLBP	7884	chr:4:1694526-1714029
SLC26A1	10861	chr:4:972860-987223
SPON2	10417	chr:4:1160719-1166979
TACC3	10460	chr:4:1723265-1746897
TMEM129	92305	chr:4:1717678-1723083
TMEM175	84286	chr:4:926261-952443
WHSC1	7468	chr:4:1873122-1983933
WHSC2	7469	chr:4:1984442-2010958
ZNF141	7700	chr:4:331595-367690
ZNF595	152687	chr:4:53226-88098
ZNF718	255403	chr:4:124419-156490
ZNF721	170960	chr:4:433776-493441

Table S40: Genes located in the interval for Wolf Hirschhorn Syndrome [MIM:194190].

S4.21 15q26 Overgrowth Syndrome [DECIPHER:81]

Term name	Term ID	Frequency	Reference
Broad nasal bridge	HP:0000431	100.00 %	46
Long philtrum	HP:0000343	100.00 %	46
High anterior hairline	HP:0009890	96.15 %	47
Abnormality of the pinna	HP:0000377	90.00 %	48
Intellectual disability	HP:0001249	90.00 %	47,48
Long face	HP:0000276	90.00 %	48 47,49
Neurological speech impairment	HP:0002167	90.00 %	49
Prominent nose	HP:0000448	86.11 %	48 47,49
Micrognathia	HP:0000347	80.00 %	46
Macrocephaly	HP:0000256	80.00 %	48
Low-set ears	HP:0000369	80.00 %	48
Triangular facies	HP:0000325	80.00 %	48
Mandibular prognathia	HP:0000303	78.79 %	47,49
Overgrowth	HP:0001548	75.00 %	47,48
Downward slanting palpebral fissures	HP:0000494	60.00 %	46 48
Bulbous nose	HP:0000414	60.00 %	46
High-arched palate	HP:0000156	60.00 %	46
Sensorineural hearing impairment	HP:0000407	60.00 %	46
Facial asymmetry	HP:0000324	53.33 %	46 48
Puffy cheeks	HP:0002262	53.33 %	49
Cardiac malformation	HP:0002564	40.00 %	46
Short neck	HP:0000470	40.00 %	48
Joint hypermobility	HP:0001382	30.00 %	48
Arachnodactyly	HP:0001166	29.03 %	46 48,49
Cardiac malformation	HP:0002564	25.64 %	48 47,49
Hydronephrosis	HP:0000126	25.00 %	47
Craniosynostosis	HP:0001363	21.43 %	47
Scoliosis	HP:0002650	20.00 %	48 47,50
Horseshoe kidney	HP:0000085	18.18 %	47,49
Tapered fingers	HP:0001182	15.38 %	48,49
Camptodactyly (hands)	HP:0100490	15.38 %	48,49
Brachydactyly (hand)	HP:0100667	11.54 %	48,49
Strabismus	HP:0000486	10.71 %	47
Renal agenesis	HP:0000104	6.25 %	47
Vesicoureteral reflux	HP:0000076	6.25 %	47
Polycystic kidney dysplasia	HP:0000113	6.25 %	47
Duplication of renal pelvis	HP:0005580	6.25 %	47

Table S41: Phenotypic annotations for 15q26 Overgrowth Syndrome [DECIPHER:81].

Gene Symbol	Entrez ID	Position
ADAMTS17	170691	chr:15:100511642-100882182
ALDH1A3	220	chr:15:101420008-101456830
ASB7	140460	chr:15:101142754-101191905

Gene Symbol	Entrez ID	Position
CHSY1	22856	chr:15:101715927-101792136
FAM138E	100124412	chr:15:102495087-102496557
IGF1R	3480	chr:15:99192760-99507758
LASS3	204219	chr:15:100940599-101084924
LINS	55180	chr:15:101109434-101142403
LRRK28	123355	chr:15:99791651-99926497
LRRK1	79705	chr:15:101459459-101610316
LYSMD4	145748	chr:15:100267609-100273625
MEF2A	4205	chr:15:100106132-100256629
OR4F15	390649	chr:15:102358389-102359327
OR4F4	26682	chr:15:102462344-102463261
OR4F6	390648	chr:15:102345922-102346860
PCSK6	5046	chr:15:101844132-102030186
PGPEP1L	145814	chr:15:99511458-99551023
SNRPA1	6627	chr:15:101821714-101835459
SYNM	23336	chr:15:99645285-99675799
TARSL2	123283	chr:15:102193954-102264644
TM2D3	80213	chr:15:102182048-102192593
TTC23	64927	chr:15:99676527-99789814

Table S42: Genes located in the interval for 15q26 Overgrowth Syndrome [DECIPHER:81].

S4.22 Chromosome 15q24 Deletion Syndrome [MIM:613406]

Term name	Term ID	Frequency	Reference
Sporadic	HP:0003745	100.00 %	51
Intellectual disability	HP:0001249	100.00 %	51,51,52
High forehead	HP:0000348	90.91 %	52
Impaired language development	HP:0000750	76.92 %	51,52
Abnormality of the outer ear	HP:0000356	70.59 %	51,52
High anterior hairline	HP:0009890	69.23 %	51
Muscular hypotonia	HP:0001252	64.71 %	51,52
Joint laxity	HP:0001388	52.94 %	51,52
Recurrent infections	HP:0002719	50.00 %	51,52
Hypertelorism	HP:0000316	47.06 %	51,52
Epicanthus	HP:0000286	47.06 %	51,52
Downward slanting palpebral fissures	HP:0000494	47.06 %	51,52
Long, smooth philtrum	HP:0000299	47.06 %	51,52
Hypospadias	HP:0000047	42.86 %	51,52
Medial flaring of the eyebrow	HP:0010747	41.18 %	51,52
Strabismus	HP:0000486	41.18 %	51,52
Long, narrow facies	HP:0000318	38.46 %	51
Cardiac malformation	HP:0002564	37.50 %	52
Micropenis	HP:0000054	35.71 %	51,52
Low birth weight	HP:0001518	35.29 %	51,52
Thick lower lip vermillion	HP:0000179	35.29 %	51,52
Conspicuously happy disposition	HP:0100024	30.77 %	51
Facial asymmetry	HP:0000324	30.77 %	51
Abnormality of the voice	HP:0001608	30.77 %	51
Autism	HP:0000717	28.57 %	51
Scoliosis	HP:0002650	27.78 %	51,52
Broad nasal root	HP:0000424	23.53 %	51
Small mouth	HP:0000160	23.53 %	51,52
Transverse palmar creases	HP:0000954	23.53 %	51,52
Inguinal hernia	HP:0000023	23.53 %	51,52
Hyperactivity	HP:0000752	23.08 %	51
Aggressive behavior	HP:0000718	23.08 %	51
Obesity	HP:0001513	23.08 %	51
Flared nostrils	HP:0000454	23.08 %	51
Radial deviation of fingers	HP:0009466	23.08 %	51
Feeding difficulties	HP:0002022	23.08 %	51
Cryptorchidism	HP:0000028	21.43 %	51,52
Diaphragmatic hernia	HP:0000776	21.43 %	51
Hearing impairment	HP:0000365	20.00 %	51,52
Microcephaly	HP:0000252	17.65 %	51,52
Widely spaced teeth	HP:0000687	17.65 %	51,52
Abnormality of the nasal bridge	HP:0000422	17.65 %	51,52
High-arched palate	HP:0000156	17.65 %	51,52
Nystagmus	HP:0000639	17.65 %	51,52
Toe syndactyly	HP:0001770	17.65 %	51,52
Proximally placed thumb	HP:0009623	17.65 %	51,52
Gastrointestinal atresia	HP:0002589	17.65 %	51,52
Dysplastic corpus callosum	HP:0006989	16.67 %	51,52
Developmental regression	HP:0002376	15.38 %	51

Term name	Term ID	Frequency	Reference
Sleep disturbances	HP:0002360	15.38 %	51
Deeply set eye	HP:0000490	15.38 %	51
Hypoplastic nasal alae	HP:0000430	15.38 %	51
Arachnodactyly	HP:0001166	15.38 %	51
Brachydactyly	HP:0001156	15.38 %	51
Hypoplastic hand	HP:0004279	15.38 %	51
Growth hormone deficiency	HP:0000824	15.38 %	51
Cafe-au-lait spots	HP:0000957	15.38 %	51
Thin lips	HP:0000213	11.76 %	51,52
Nares, anteverted	HP:0000463	11.76 %	51,52
Hypermetropia	HP:0000540	11.76 %	51,52
Polyhydramnios	HP:0001561	7.69 %	51
Up-slanting palpebral fissures	HP:0000582	5.88 %	51,52
Microphthalmos	HP:0000568	5.88 %	51,52
Anisocoria	HP:0009916	5.88 %	51,52
Iris coloboma	HP:0000612	5.88 %	51,52

Table S43: Phenotypic annotations for Chromosome 15q24 Deletion Syndrome [MIM:613406].

Gene Symbol	Entrez ID	Position
ARID3B	10620	chr:15:74833547-74890471
C15orf17	57184	chr:15:75192327-75199461
C15orf39	56905	chr:15:75494220-75504509
CCDC33	80125	chr:15:74528666-74628481
CLK3	1198	chr:15:74900712-74922541
COMMD4	54939	chr:15:75628373-75632613
COX5A	9377	chr:15:75212615-75230494
CPLX3	594855	chr:15:75118950-75124135
CSK	1445	chr:15:75074424-75095538
CSPG4	1464	chr:15:75966662-76005188
CYP11A1	1583	chr:15:74630102-74660080
CYP1A1	1543	chr:15:75011882-75017876
CYP1A2	1544	chr:15:75041183-75048940
EDC3	80153	chr:15:74922898-74988385
IMP3	55272	chr:15:75931425-75932663
ISLR	3671	chr:15:74466086-74469211
ISLR2	57611	chr:15:74421714-74429142
LMAN1L	79748	chr:15:75105193-75118098
MAN2C1	4123	chr:15:75648132-75660940
MPI	4351	chr:15:75182409-75190564
NEIL1	79661	chr:15:75639330-75647587
ODF3L1	161753	chr:15:76016318-76020026
PPCDC	60490	chr:15:75315926-75343066
PTPN9	5780	chr:15:75759461-75871624
RPP25	54913	chr:15:75247442-75249774
SCAMP2	10066	chr:15:75137196-75165669
SCAMP5	192683	chr:15:75287875-75313835

Gene Symbol	Entrez ID	Position
SEMA7A	8482	chr:15:74701629-74726298
SIN3A	25942	chr:15:75661719-75748123
SNUPN	10073	chr:15:75890423-75918718
SNX33	257364	chr:15:75941347-75950967
STRA6	64220	chr:15:74471806-74501370
UBE2Q2	92912	chr:15:76135621-76193387
UBL7	84993	chr:15:74738317-74753528
ULK3	25989	chr:15:75128458-75135551
GOLGA6A	342096	chr:15:74362197-74374890

Table **S44**: Genes located in the interval for Chromosome 15q24 Deletion Syndrome [MIM:613406].

S4.23 Smith Magenis Syndrome [MIM:182290]

Term name	Term ID	Frequency	Ref.
Isolated cases	HP:0001420	100.00 %	53
Autosomal dominant inheritance	HP:0000006	100.00 %	53
Brachycephaly	HP:0000248	75.00 %	53
Malar hypoplasia	HP:0000272	75.00 %	53
Broad face	HP:0000283	75.00 %	53
Brachydactyly	HP:0001156	75.00 %	53
Peripheral neuropathy	HP:0009830	75.00 %	53
Hyporeflexia	HP:0001265	75.00 %	53
Hoarse voice	HP:0001609	75.00 %	53
Sleep disturbances	HP:0002360	75.00 %	53
Mandibular prognathia	HP:0000303	75.00 %	53
Everted upper lip vermillion	HP:0010803	75.00 %	53
Deeply set eye	HP:0000490	75.00 %	53
Hypoplastic hand	HP:0004279	75.00 %	53
Broad hands	HP:0001169	75.00 %	53
Abnormality of the teeth	HP:0000164	75.00 %	53
Middle ear malformations	HP:0008609	75.00 %	53
Stereotyped, repetitive behaviour	HP:0000733	75.00 %	53
Generalized hypotonia	HP:0001290	75.00 %	53
Self-mutilation	HP:0000742	75.00 %	53
Speech delay	HP:0002117	75.00 %	53
Hearing impairment	HP:0000365	50.00 %	53
Broad nasal bridge	HP:0000431	50.00 %	2
Hyperactivity	HP:0000752	50.00 %	1
Scoliosis	HP:0002650	50.00 %	53
Short stature	HP:0004322	50.00 %	53
Ventriculomegaly	HP:0002119	50.00 %	53
Hyperacusis	HP:0010780	50.00 %	53
Abnormality of the tracheobronchial system	HP:0005607	50.00 %	53
Velopharyngeal insufficiency	HP:0000220	50.00 %	53
Hypercholesterolemia	HP:0003124	50.00 %	53
Hypertriglyceridemia	HP:0002155	50.00 %	53
Constipation	HP:0002019	50.00 %	53
EEG abnormalities	HP:0002353	50.00 %	53
Synophrys	HP:0000664	50.00 %	2
Abnormality of the outer ear	HP:0000356	50.00 %	2
Pes planus	HP:0001763	50.00 %	2
Decreased pain sensation	HP:0007328	50.00 %	2
Cardiac malformation	HP:0002564	33.00 %	53
Seizures	HP:0001250	33.00 %	53
Abnormality of the thyroid gland	HP:0000820	33.00 %	53
Abnormality of the immune system	HP:0002715	33.00 %	53
Abnormality of the urinary system	HP:0000079	7.50 %	53
Abnormality of the forearm	HP:0002973	7.50 %	53
Cleft lip/palate	HP:0000202	7.50 %	53
Retinal detachment	HP:0000541	7.50 %	53

Term name	Term ID	Frequency	Ref.
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Table S45: Phenotypic annotations for Smith Magenis Syndrome [MIM:182290].

Gene Symbol	Entrez ID	Position
TNFRSF13B	23495	chr:17:16842397-16875401
FLCN	201163	chr:17:17115525-17140501
COPS3	8533	chr:17:17150138-17184590
NT5M	56953	chr:17:17206679-17250976
MED9	55090	chr:17:17380299-17396533
RASD1	51655	chr:17:17397750-17399706
PEMT	10400	chr:17:17408876-17494993
RAII	10743	chr:17:17584786-17714766
SREBF1	6720	chr:17:17714662-17740324
TOM1L2	146691	chr:17:17746821-17875783
LRRC48	83450	chr:17:17876126-17920202
ATPAF2	91647	chr:17:17921333-17942479
C17orf39	79018	chr:17:17942610-17971717
DRG2	1819	chr:17:17991282-18011291
MYO15A	51168	chr:17:18012019-18083115
ALKBH5	54890	chr:17:18086866-18113267
LLGL1	3996	chr:17:18128935-18148188
FLII	2314	chr:17:18148149-18162054
SMCR7	125170	chr:17:18163847-18169094
TOP3A	7156	chr:17:18177234-18218320
SMCR8	140775	chr:17:18218593-18231369
SHMT1	6470	chr:17:18231186-18266855
TBC1D28	254272	chr:17:18538841-18547739
TRIM16L	147166	chr:17:18625401-18639430
FBXW10	10517	chr:17:18647325-18682661
FAM18B1	51030	chr:17:18684581-18710025
PRPSAP2	5636	chr:17:18761491-18834579
SLC5A10	125206	chr:17:18855477-18924002
FAM83G	644815	chr:17:18874380-18908059
GRAP	10750	chr:17:18923989-18950335
EPN2	22905	chr:17:19140689-19240027
B9D1	27077	chr:17:19246482-19266045
MAPK7	5598	chr:17:19281033-19286856
MFAP4	4239	chr:17:19286754-19290492
SLC47A1	55244	chr:17:19437166-19482345
ALDH3A2	224	chr:17:19552063-19580907
SLC47A2	146802	chr:17:19581627-19620042
ALDH3A1	218	chr:17:19641296-19651745
ULK2	9706	chr:17:19674142-19771238
AKAP10	11216	chr:17:19808748-19881128
SPECC1	92521	chr:17:19990334-20218067
SMCR5	140771	chr:17:17679999-17682842
SNORA59B	677882	chr:17:19460872-19461023

Gene Symbol	Entrez ID	Position
SNORD3B-1	26851	chr:17:18965224-18965440

Table **S46**: Genes located in the interval for Smith Magenis Syndrome [MIM:182290].

S4.24 Chromosome 17q11.2 deletion syndrome [MIM:613675]

Also known as neurofibromatosis I microdeletion syndrome.

Term name	Term ID	Frequency	Reference
Sporadic	HP:0003745	100.00 %	
Axillary freckling	HP:0000997	96.55 %	54
Cafe-au-lait spots	HP:0000957	93.10 %	54
Lisch nodules	HP:0009737	93.10 %	54
Hypertelorism	HP:0000316	86.21 %	54
Plexiform neurofibroma	HP:0009732	75.86 %	54
Subcutaneous neurofibromas	HP:0100698	75.86 %	54
Joint hypermobility	HP:0001382	72.41 %	54
Spinal neurofibromas	HP:0009735	64.29 %	54
Coarse facial features	HP:0000280	58.62 %	54
Bone cysts	HP:0100696	50.00 %	54
Speech difficulties	HP:0006936	48.28 %	54
Tall stature	HP:0000098	46.43 %	54
Large hands and feet	HP:0002820	46.43 %	54
Learning disability	HP:0001328	44.83 %	54
Muscular hypotonia	HP:0001252	44.83 %	54
Hyperintense lesions in the basal ganglia on MRI	HP:0007183	44.83 %	54
Scoliosis	HP:0002650	42.86 %	54
Macrocephaly	HP:0000256	39.13 %	54
Attention deficit hyperactivity disorder	HP:0007018	33.33 %	54
Broad neck	HP:0000475	31.03 %	54
Pectus excavatum	HP:0000767	31.03 %	54
Cardiac malformation	HP:0002564	28.57 %	54
Facial asymmetry	HP:0000324	27.59 %	54
Neurofibrosarcoma	HP:0100697	20.69 %	54
Optic glioma	HP:0009734	18.52 %	54
Pes cavus	HP:0001761	17.24 %	54
Malformation of the central nervous system	HP:0007319	17.24 %	54
Strabismus	HP:0000486	13.79 %	54
Low-set ears	HP:0000369	13.79 %	54
Hearing impairment	HP:0000365	10.34 %	54
Epilepsy	HP:0001275	6.90 %	54

Table S47: Phenotypic annotations for Chromosome 17q11.2 deletion syndrome [MIM:613675]

Gene Symbol	Entrez ID	Position
ADAP2	55803	chr:17:29248753-29286210
ATAD5	79915	chr:17:29159019-29222294
C17orf42	79736	chr:17:29226000-29233285
EVI2A	2123	chr:17:29643427-29648766

Gene Symbol	Entrez ID	Position
EVI2B	2124	chr:17:29630787-29641129
MIR193A	406968	chr:17:29887014-29887101
MIR365-2	100126356	chr:17:29902429-29902539
NF1	4763	chr:17:29421944-29704694
OMG	4974	chr:17:29621667-29624379
RAB11FIP4	84440	chr:17:29718641-29865231
RNF135	84282	chr:17:29297955-29326928
UTP6	55813	chr:17:30190189-30228728

Table **S48**: Genes located in the interval for Chromosome 17q11.2 deletion syndrome [MIM:613675]

S4.25 Xq28 (MECP2) Duplication [DECIPHER:45]

Term name	Term ID	Frequency	Reference
X-linked recessive inheritance	HP:0001419	100.00 %	55
Intellectual disability, severe	HP:0010864	100.00 %	55,56
Dysphagia	HP:0002015	87.50 %	56
Gastroesophageal reflux	HP:0002020	81.25 %	56
Recurrent respiratory infections	HP:0002205	75.00 %	55
Spasticity, progressive	HP:0002191	70.00 %	56
Absent speech development	HP:0001344	70.00 %	55
Drooling	HP:0002307	66.67 %	56
Neonatal hypotonia	HP:0001319	50.00 %	55
Seizures	HP:0001250	50.00 %	55
Feeding difficulties	HP:0002022	50.00 %	55
Failure to thrive	HP:0001508	50.00 %	55
Early death	HP:0001432	50.00 %	55
Decreased IgA	HP:0002720	40.00 %	56
Inability to walk	HP:0002540	33.00 %	55
Small mouth	HP:0000160	22.22 %	56
Microcephaly	HP:0000252	12.90 %	56
Autism	HP:0000717	7.50 %	55
Brachycephaly	HP:0000248	7.50 %	55
Malar hypoplasia	HP:0000272	7.50 %	55
Large ears	HP:0000400	7.50 %	55
Flattened nasal bridge	HP:0000425	7.50 %	55
Gait ataxia	HP:0002066	7.50 %	55
Constipation	HP:0002019	7.50 %	55
Hypoplasia of the corpus callosum	HP:0002079	7.50 %	55
Functional abnormality of the bladder	HP:0000009	7.50 %	55
Anxiety	HP:0000739	7.50 %	55
Stereotypical motor behaviors	HP:0008758	7.50 %	55
Autism	HP:0000717	5.00 %	55
Depression	HP:0000716	5.00 %	55
Anxiety	HP:0000739	5.00 %	55

Table S49: Phenotypic annotations for Xq28 (MECP2) Duplication [DECIPHER:45]

Gene Symbol	Entrez ID	Position
ABCD1	215	chr:X:152990322-153010215
NAA10	8260	chr:X:153195376-153200467
ARHGAP4	393	chr:X:153172829-153191713
ATP2B3	492	chr:X:152801579-152848386
AVPR2	554	chr:X:153167984-153172619
BCAP31	10134	chr:X:152965946-152990200
BGN	633	chr:X:152760346-152775003
DUSP9	1852	chr:X:152907896-152916780
FAM58A	92002	chr:X:152853382-152864631

Gene Symbol	Entrez ID	Position
HCFC1	3054	chr:X:153213007-153236818
IDH3G	3421	chr:X:153051220-153059966
IRAK1	3654	chr:X:153275956-153285341
L1CAM	3897	chr:X:153126970-153141398
MECP2	4204	chr:X:153287263-153363187
PDZD4	57595	chr:X:153067622-153096002
PLXNB3	5365	chr:X:153029650-153044800
PNCK	139728	chr:X:152935187-152939815
RENBP	5973	chr:X:153200721-153210231
SLC6A8	6535	chr:X:152953751-152962047
SRPK3	26576	chr:X:153046455-153051186
SSR4	6748	chr:X:153060093-153063953
TMEM187	8269	chr:X:153237990-153248645
HAUS7	5559	chr:X:152713122-152736602

Table **S50**: Genes located in the interval for Xq28 (MECP2) Duplication [DECIPHER:45]

S4.26 Chromosome 17q21.31 Microdeletion Syndrome [MIM:610443]

CHROMOSOME 17Q21.31 MICRODELETION SYNDROME (MIM:610443)

Term name	Term ID	Frequency	Reference
Sporadic	HP:0003745	100.00 %	57
Autosomal dominant inheritance	HP:0000006	100.00 %	57
Variable expressivity	HP:0003828	100.00 %	57
Generalized hypotonia	HP:0001290	90.00 %	57
Joint hypermobility	HP:0001382	75.00 %	57
Abnormality of hair pigmentation	HP:0009887	75.00 %	57
Abnormality of hair texture	HP:0010719	75.00 %	57
Nasal speech	HP:0001611	75.00 %	57
Narrow palate	HP:0000189	75.00 %	57
High palate	HP:0000218	75.00 %	57
Slender fingers	HP:0001238	75.00 %	57
Malformation of the central nervous system	HP:0007319	75.00 %	57
Feeding difficulties	HP:0002022	75.00 %	57
Abnormality of the teeth	HP:0000164	75.00 %	57
Prominent fingertip pads	HP:0001212	75.00 %	57
Cryptorchidism	HP:0000028	71.00 %	57
Epilepsy	HP:0001275	55.00 %	57
Speech delay	HP:0002117	50.00 %	57
Broad forehead	HP:0000337	50.00 %	57
High forehead	HP:0000348	50.00 %	57
Ptosis	HP:0000508	50.00 %	57
Blepharophimosis	HP:0000581	50.00 %	57
Upstalting palpebral fissures	HP:0000582	50.00 %	57
Epicanthus	HP:0000286	50.00 %	57
Large ears	HP:0000400	50.00 %	57
Pear-shaped nose	HP:0000447	50.00 %	57
Bulbous nasal tip	HP:0000443	50.00 %	57
Everted lower lip vermillion	HP:0000232	50.00 %	57
Conspicuously happy disposition	HP:0100024	50.00 %	57
Cardiac malformation	HP:0002564	39.00 %	57
Abnormality of the urinary system	HP:0000079	37.00 %	57
Hypermetropia	HP:0000540	33.00 %	57
Strabismus	HP:0000486	33.00 %	57
Narrow hand	HP:0004283	33.00 %	57
Hypotrophy of the small hand muscles	HP:0006006	33.00 %	57
Hip dysplasia	HP:0001385	33.00 %	57
Dislocated hips	HP:0002827	33.00 %	57
Positional foot deformities	HP:0005656	33.00 %	57
Scoliosis	HP:0002650	33.00 %	57
Kyphosis	HP:0002808	33.00 %	57
Low birth weight	HP:0001518	7.50 %	57
Short stature	HP:0004322	7.50 %	57
Pectus excavatum	HP:0000767	7.50 %	57
Ridging of metopic suture	HP:0005751	5.00 %	57
Cleft palate	HP:0000175	5.00 %	57
Microcephaly	HP:0000252	5.00 %	57
Cataract	HP:0000518	5.00 %	57

CHROMOSOME 17Q21.31 MICRODELETION SYNDROME (MIM:610443)

Term name	Term ID	Frequency	Reference
Pyloric stenosis	HP:0002021	5.00 %	⁵⁷
Vertebral fusion	HP:0002948	5.00 %	⁵⁷
Spondylolisthesis	HP:0003302	5.00 %	⁵⁷
Hypotelorism	HP:0000601	5.00 %	⁵⁷

Table S51: Phenotypic annotations for Chromosome 17q21.31 Microdeletion Syndrome [MIM:610443].

Gene Symbol	Entrez ID	Position
C17orf69	147081	chr:17:43716340-43723594
CRHR1	1394	chr:17:43861645-43913193
KIAA1267	284058	chr:17:44107281-44302739
MAPT	4137	chr:17:43971747-44105699
STH	246744	chr:17:44076615-44077059
IMP5	162540	chr:17:43922255-43924437

Table S52: Genes located in the interval for Chromosome 17q21.31 Microdeletion Syndrome [MIM:610443].

S4.27 Adenomatous polyposis of the colon [MIM:175100]

Term name	Term ID	Frequency	Reference
Autosomal dominant inheritance	HP:0000006	100.00 %	1
Variable expressivity	HP:0003828	100.00 %	1
Multiple adenomatous colon polyps	HP:0005227	90.00 %	1
multiple duodenal polyps	HP:0004783	70.00 %	58
Colon cancer	HP:0003003	33.00 %	1
Multiple gastric polyps	HP:0004394	33.00 %	1
Adrenocortical adenoma	HP:0008256	13.00 %	59
Desmoid tumors	HP:0100245	10.00 %	58
Duodenal carcinoma	HP:0006771	8.00 %	58
Carious teeth	HP:0000670	7.50 %	1
Increased number of teeth	HP:0011069	7.50 %	1
Unerupted tooth	HP:0000706	7.50 %	1
Fibroma of the breast	HP:0010619	7.50 %	1
Congenital hypertrophy of retinal pigment epithelium	HP:0007649	7.50 %	1
Skin cysts	HP:0200040	7.50 %	60,61
Osteoma	HP:0100246	7.50 %	60,61
Hepatoblastoma	HP:0002884	1.60 %	58
Papillary thyroid carcinoma	HP:0002895	1.50 %	58
Medulloblastoma	HP:0002885	1.00 %	58

Table S53: Phenotypic annotations for Adenomatous polyposis of the colon [MIM:175100].

Gene Symbol	Entrez ID	Position
APC	324	chr:5:112043217-112181935
SRP19	6728	chr:5:112196992-112203603
REEP5	7905	chr:5:112212080-112258030

Table S54: Genes located in the interval for Adenomatous polyposis of the colon [MIM:175100].

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